

BeGECS panel		
versie	v2 (1704 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>AAAS</i>	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
<i>AARS2</i>	612035	Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive
<i>ABAT</i>	137150	GABA-transaminase deficiency, 613163 (3), Autosomal recessive
<i>ABCA1</i>	600046	Tangier disease, 205400 (3), Autosomal recessive; HDL deficiency, familial, 1, 604091 (3), Autosomal dominant
<i>ABCA12</i>	607800	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive
<i>ABCA3</i>	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
<i>ABCA4</i>	601691	Retinal dystrophy, early-onset severe, 248200 (3), Autosomal recessive; Retinitis pigmentosa 19, 601718 (3), Autosomal recessive; {Macular degeneration, age-related, 2}, 153800 (3), Autosomal dominant; Cone-rod dystrophy 3, 604116 (3), Autosomal recessive; Fundus flavimaculatus, 248200 (3), Autosomal recessive; Stargardt disease 1, 248200 (3), Autosomal recessive
<i>ABCB11</i>	603201	Cholestasis, benign recurrent intrahepatic, 2, 605479 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 2, 601847 (3), Autosomal recessive
<i>ABCB4</i>	171060	Gallbladder disease 1, 600803 (3), Autosomal dominant, Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 3, 614972 (3), Autosomal dominant, Autosomal recessive; Cholestasis, progressive familial intrahepatic 3, 602347 (3), Autosomal recessive
<i>ABCB7</i>	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive
<i>ABCC2</i>	601107	Dubin-Johnson syndrome, 237500 (3), Autosomal recessive
<i>ABCC6</i>	603234	Pseudoxanthoma elasticum, 264800 (3), Autosomal recessive; Arterial calcification, generalized, of infancy, 2, 614473 (3), Autosomal recessive; Pseudoxanthoma elasticum, forme fruste, 177850 (3), Autosomal dominant

<i>ABCC8</i>	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal dominant, Autosomal recessive
<i>ABCD1</i>	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<i>ABCD3</i>	170995	?Bile acid synthesis defect, congenital, 5, 616278 (3), Autosomal recessive
<i>ABCD4</i>	603214	Methylmalonic aciduria and homocystinuria, cblJ type, 614857 (3), Autosomal recessive
<i>ABCG5</i>	605459	Sitosterolemia 2, 618666 (3), Autosomal recessive
<i>ABCG8</i>	605460	Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3)
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ABHD5</i>	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
<i>ACAD9</i>	611103	Mitochondrial complex I deficiency, nuclear type 20, 611126 (3), Autosomal recessive
<i>ACADM</i>	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3), Autosomal recessive
<i>ACADS</i>	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3), Autosomal recessive
<i>ACADSB</i>	600301	2-methylbutyrylglycinuria, 610006 (3), Autosomal recessive
<i>ACADVL</i>	609575	VLCAD deficiency, 201475 (3), Autosomal recessive
<i>ACAT1</i>	607809	Alpha-methylacetoacetic aciduria, 203750 (3), Autosomal recessive
<i>ACE</i>	106180	{Stroke, hemorrhagic}, 614519 (3); Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Myocardial infarction, susceptibility to} (3); {Microvascular complications of diabetes 3}, 612624 (3); [Angiotensin I-converting enzyme, benign serum increase] (3); {SARS, progression of} (3)
<i>ACO2</i>	100850	?Optic atrophy 9, 616289 (3), Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
<i>ACOX1</i>	609751	Mitchell syndrome, 618960 (3), Autosomal dominant; Peroxisomal acyl-CoA oxidase deficiency, 264470 (3), Autosomal recessive
<i>ACP5</i>	171640	Spondyloenchondrodysplasia with immune dysregulation, 607944 (3), Autosomal recessive
<i>ACSF3</i>	614245	Combined malonic and methylmalonic aciduria, 614265 (3)
<i>ACSL4</i>	300157	Intellectual developmental disorder, X-linked 63, 300387 (3), X-linked dominant

<i>ACTA1</i>	102610	?Myopathy, scapulohumeroperoneal, 616852 (3), Autosomal dominant; Nemaline myopathy 3, autosomal dominant or recessive, 161800 (3), Autosomal dominant, Autosomal recessive; Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3), Autosomal dominant, Autosomal recessive; Myopathy, actin, congenital, with cores, 161800 (3), Autosomal dominant, Autosomal recessive; Myopathy, congenital, with fiber-type disproportion 1, 255310 (3), Autosomal dominant, Autosomal recessive
<i>ACY1</i>	104620	Aminoacylase 1 deficiency, 609924 (3), Autosomal recessive
<i>ADA</i>	608958	Adenosine deaminase deficiency, partial, 102700 (3), Somatic mosaicism, Autosomal recessive; Severe combined immunodeficiency due to ADA deficiency, 102700 (3), Somatic mosaicism, Autosomal recessive
<i>ADA2</i>	607575	Sneddon syndrome, 182410 (3), Autosomal recessive; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 (3), Autosomal recessive
<i>ADAMTS10</i>	608990	Weill-Marchesani syndrome 1, recessive, 277600 (3), Autosomal recessive
<i>ADAMTS13</i>	604134	Thrombotic thrombocytopenic purpura, hereditary, 274150 (3), Autosomal recessive
<i>ADAMTS17</i>	607511	Weill-Marchesani 4 syndrome, recessive, 613195 (3), Autosomal recessive
<i>ADAMTS2</i>	604539	Ehlers-Danlos syndrome, dermatosparaxis type, 225410 (3), Autosomal recessive
<i>ADAMTSL2</i>	612277	Geleophysic dysplasia 1, 231050 (3), Autosomal recessive
<i>ADAR</i>	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<i>ADAT3</i>	615302	Neurodevelopmental disorder with brain abnormalities, poor growth, and dysmorphic facies, 615286 (3), Autosomal recessive
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<i>ADGRV1</i>	602851	Usher syndrome, type 2C, 605472 (3), Digenic dominant, Autosomal recessive; Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 (3), Digenic dominant, Autosomal recessive; ?Febrile seizures, familial, 4, 604352 (3), Autosomal dominant
<i>ADK</i>	102750	Hypermethioninemia due to adenosine kinase deficiency, 614300 (3), Autosomal recessive
<i>ADSL</i>	608222	Adenylosuccinase deficiency, 103050 (3), Autosomal recessive
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AGA</i>	613228	Aspartylglucosaminuria, 208400 (3), Autosomal recessive
<i>AGBL5</i>	615900	Retinitis pigmentosa 75, 617023 (3), Autosomal recessive

<i>AGK</i>	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
<i>AGL</i>	610860	Glycogen storage disease IIIa, 232400 (3), Autosomal recessive; Glycogen storage disease IIIb, 232400 (3), Autosomal recessive
<i>AGPAT2</i>	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
<i>AGPS</i>	603051	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3), Autosomal recessive
<i>AGRN</i>	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
<i>AGT</i>	106150	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; {Preeclampsia, susceptibility to} (3); {Hypertension, essential, susceptibility to}, 145500 (3), Multifactorial
<i>AGTR1</i>	106165	{Hypertension, essential}, 145500 (3), Multifactorial; Renal tubular dysgenesis, 267430 (3), Autosomal recessive
<i>AGXT</i>	604285	Hyperoxaluria, primary, type 1, 259900 (3), Autosomal recessive
<i>AHCY</i>	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752 (3), Autosomal recessive
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>AICDA</i>	605257	Immunodeficiency with hyper-IgM, type 2, 605258 (3), Autosomal recessive
<i>AIFM1</i>	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
<i>AIMP1</i>	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<i>AIPL1</i>	604392	Leber congenital amaurosis 4, 604393 (3), Autosomal dominant, Autosomal recessive; Retinitis pigmentosa, juvenile, 604393 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy, 604393 (3), Autosomal dominant, Autosomal recessive
<i>AIRE</i>	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal dominant, Autosomal recessive
<i>AK1</i>	103000	Hemolytic anemia due to adenylate kinase deficiency, 612631 (3), Autosomal recessive
<i>AK2</i>	103020	Reticular dysgenesis, 267500 (3), Autosomal recessive
<i>AKR1C2</i>	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
<i>AKR1D1</i>	604741	Bile acid synthesis defect, congenital, 2, 235555 (3), Autosomal recessive
<i>ALAD</i>	125270	Porphyria, acute hepatic, 612740 (3), Autosomal recessive; {Lead poisoning, susceptibility to}, 612740 (3), Autosomal recessive
<i>ALAS2</i>	301300	Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyrin, erythropoietic, X-linked, 300752 (3), X-linked

<i>ALDH18A1</i>	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
<i>ALDH1A3</i>	600463	Microphthalmia, isolated 8, 615113 (3), Autosomal recessive
<i>ALDH3A2</i>	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<i>ALDH4A1</i>	606811	Hyperprolinemia, type II, 239510 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>ALDH7A1</i>	107323	Epilepsy, pyridoxine-dependent, 266100 (3), Autosomal recessive
<i>ALDOB</i>	612724	Fructose intolerance, hereditary, 229600 (3), Autosomal recessive
<i>ALG1</i>	605907	Congenital disorder of glycosylation, type Ik, 608540 (3), Autosomal recessive
<i>ALG11</i>	613666	Congenital disorder of glycosylation, type Ip, 613661 (3), Autosomal recessive
<i>ALG12</i>	607144	Congenital disorder of glycosylation, type Ig, 607143 (3), Autosomal recessive
<i>ALG2</i>	607905	Congenital disorder of glycosylation, type Ii, 607906 (3), Autosomal recessive; Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228 (3), Autosomal recessive
<i>ALG3</i>	608750	Congenital disorder of glycosylation, type Id, 601110 (3), Autosomal recessive
<i>ALG6</i>	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
<i>ALG8</i>	608103	Congenital disorder of glycosylation, type Ih, 608104 (3), Autosomal recessive; Polycystic liver disease 3 with or without kidney cysts, 617874 (3), Autosomal dominant
<i>ALG9</i>	606941	Gillessen-Kaesbach-Nishimura syndrome, 263210 (3), Autosomal recessive; Congenital disorder of glycosylation, type II, 608776 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ALOX12B</i>	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
<i>ALOXE3</i>	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
<i>ALPL</i>	171760	Odontohypophosphatasia, 146300 (3), Autosomal dominant, Autosomal recessive; Hypophosphatasia, infantile, 241500 (3), Autosomal recessive; Hypophosphatasia, childhood, 241510 (3), Autosomal recessive; Hypophosphatasia, adult, 146300 (3), Autosomal dominant, Autosomal recessive
<i>ALS2</i>	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal

		recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive
<i>ALX3</i>	606014	Frontonasal dysplasia 1, 136760 (3), Autosomal recessive
<i>ALX4</i>	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
<i>AMACR</i>	604489	Alpha-methylacyl-CoA racemase deficiency, 614307 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 4, 214950 (3), Autosomal recessive
<i>AMER1</i>	300647	Osteopathia striata with cranial sclerosis, 300373 (3), X-linked dominant
<i>AMH</i>	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
<i>AMHR2</i>	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
<i>AMN</i>	605799	Imerslund-Grasbeck syndrome 2, 618882 (3), Autosomal recessive
<i>AMPD2</i>	102771	?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive
<i>AMT</i>	238310	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>ANO10</i>	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<i>ANOS</i>	608662	Muscular dystrophy, limb-girdle, autosomal recessive 12, 611307 (3), Autosomal recessive; Miyoshi muscular dystrophy 3, 613319 (3), Autosomal recessive; Gnathodiaphyseal dysplasia, 166260 (3), Autosomal dominant
<i>ANOS1</i>	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
<i>ANTXR1</i>	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
<i>ANTXR2</i>	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
<i>AP1S2</i>	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
<i>AP3B1</i>	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
<i>AP3B2</i>	602166	Developmental and epileptic encephalopathy 48, 617276 (3), Autosomal recessive
<i>AP4B1</i>	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<i>AP4E1</i>	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
<i>AP4M1</i>	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<i>AP4S1</i>	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive

<i>APOB</i>	107730	Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
<i>APOC2</i>	608083	Hyperlipoproteinemia, type Ib, 207750 (3), Autosomal recessive
<i>APRT</i>	102600	Adenine phosphoribosyltransferase deficiency, 614723 (3), Autosomal recessive
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>AQP2</i>	107777	Diabetes insipidus, nephrogenic, 2, 125800 (3), Autosomal dominant, Autosomal recessive
<i>AR</i>	313700	Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation; Androgen insensitivity, 300068 (3), X-linked recessive; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive
<i>ARFGF2</i>	605371	Periventricular heterotopia with microcephaly, 608097 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ARHGEF9</i>	300429	Developmental and epileptic encephalopathy 8, 300607 (3), X-linked
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6</i>	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
<i>ARPC1B</i>	604223	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718 (3), Autosomal recessive
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ARSB</i>	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200 (3), Autosomal recessive
<i>ARSL</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ARV1</i>	611647	Developmental and epileptic encephalopathy 38, 617020 (3), Autosomal recessive
<i>ARX</i>	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
<i>ASAH1</i>	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
<i>ASCC1</i>	614215	Spinal muscular atrophy with congenital bone fractures 2, 616867 (3), Autosomal recessive; Barrett esophagus/esophageal adenocarcinoma, 614266 (3)
<i>ASL</i>	608310	Argininosuccinic aciduria, 207900 (3), Autosomal recessive

<i>ASNS</i>	108370	Asparagine synthetase deficiency, 615574 (3), Autosomal recessive
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ASPM</i>	605481	Microcephaly 5, primary, autosomal recessive, 608716 (3), Autosomal recessive
<i>ASS1</i>	603470	Citrullinemia, 215700 (3), Autosomal recessive
<i>ATAD1</i>	614452	Hyperekplexia 4, 618011 (3), Autosomal recessive
<i>ATCAY</i>	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<i>ATF6</i>	605537	Achromatopsia 7, 616517 (3), Autosomal recessive
<i>ATIC</i>	601731	AICA-ribosiduria due to ATIC deficiency, 608688 (3), Autosomal recessive
<i>ATM</i>	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
<i>ATOH7</i>	609875	Persistent hyperplastic primary vitreous, autosomal recessive, 221900 (3), Autosomal recessive
<i>ATP13A2</i>	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
<i>ATP5F1E</i>	606153	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053 (3), Autosomal recessive
<i>ATP6AP1</i>	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
<i>ATP6V0A2</i>	611716	Wrinkly skin syndrome, 278250 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIA, 219200 (3), Autosomal recessive
<i>ATP6V0A4</i>	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss, 602722 (3), Autosomal recessive
<i>ATP6V1B1</i>	192132	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss, 267300 (3), Autosomal recessive
<i>ATP7A</i>	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>ATP8A2</i>	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<i>ATP8B1</i>	602397	Cholestasis, progressive familial intrahepatic 1, 211600 (3), Autosomal recessive; Cholestasis, intrahepatic, of pregnancy, 1, 147480 (3), Autosomal dominant; Cholestasis, benign recurrent intrahepatic, 243300 (3), Autosomal recessive
<i>ATPAF2</i>	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273 (3), Autosomal recessive
<i>ATR</i>	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant

<i>ATRX</i>	300032	Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
<i>AUH</i>	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<i>B3GALNT2</i>	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181 (3), Autosomal recessive
<i>B3GALT6</i>	615291	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 (3), Autosomal recessive; Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 (3), Autosomal recessive; Al-Gazali syndrome, 609465 (3), Autosomal recessive
<i>B3GAT3</i>	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600 (3), Autosomal recessive
<i>B3GLCT</i>	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>B4GALT1</i>	137060	Congenital disorder of glycosylation, type IIc, 607091 (3), Autosomal recessive
<i>B4GALT7</i>	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 (3), Autosomal recessive
<i>B4GAT1</i>	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<i>B9D1</i>	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
<i>B9D2</i>	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
<i>BBIP1</i>	613605	?Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>BCHE</i>	177400	Butyrylcholinesterase deficiency, 617936 (3), Autosomal recessive; {Apnea, postanesthetic, susceptibility to, due to BCHE deficiency}, 617936 (3), Autosomal recessive
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BCKDK</i>	614901	Branched-chain keto acid dehydrogenase kinase deficiency, 614923 (3)
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant

<i>BCS1L</i>	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
<i>BEST1</i>	607854	Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinopathopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3)
<i>BGN</i>	301870	Meester-Loeys syndrome, 300989 (3), X-linked; Spondyloepimetaphyseal dysplasia, X-linked, 300106 (3), X-linked recessive
<i>BIN1</i>	601248	Centronuclear myopathy 2, 255200 (3), Autosomal recessive
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BLOC1S6</i>	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>BLTP1</i> (<i>KIAA1109</i>)	611565	Alkuraya-Kucinskas syndrome, 617822 (3), Autosomal recessive
<i>BMP1</i>	112264	Osteogenesis imperfecta, type XIII, 614856 (3), Autosomal recessive
<i>BMPER</i>	608699	Diaphanospondylodysostosis, 608022 (3), Autosomal recessive
<i>BMPR1B</i>	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
<i>BOLA3</i>	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299 (3), Autosomal recessive
<i>BRAT1</i>	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 (3), Autosomal recessive; Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 (3), Autosomal recessive
<i>BRCA2</i>	600185	Fanconi anemia, complementation group D1, 605724 (3), Autosomal recessive; {Glioblastoma 3}, 613029 (3), Autosomal recessive; {Medulloblastoma}, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive; {Prostate cancer}, 176807 (3), Autosomal dominant, Somatic mutation; {Breast-ovarian cancer, familial, 2}, 612555 (3), Autosomal dominant; {Breast cancer, male, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; {Pancreatic cancer 2}, 613347 (3); Wilms tumor, 194070 (3), Autosomal dominant, Somatic mutation
<i>BRF1</i>	604902	Cerebellofaciodental syndrome, 616202 (3), Autosomal recessive
<i>BRIP1</i>	605882	Fanconi anemia, complementation group J, 609054 (3); {Breast cancer, early-onset, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation
<i>BRWD3</i>	300553	Intellectual developmental disorder, X-linked 93, 300659 (3), X-linked recessive

<i>BSCL2</i>	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VC, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
<i>BSND</i>	606412	Sensorineural deafness with mild renal dysfunction, 602522 (3), Autosomal recessive; Bartter syndrome, type 4a, 602522 (3), Autosomal recessive
<i>BTB</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>BTK</i>	300300	Agammaglobulinemia, X-linked 1, 300755 (3), X-linked recessive; Isolated growth hormone deficiency, type III, with agammaglobulinemia, 307200 (3), X-linked recessive
<i>BUB1B</i>	602860	Colorectal cancer, somatic, 114500 (3); [Premature chromatid separation trait], 176430 (3), Autosomal dominant; Mosaic variegated aneuploidy syndrome 1, 257300 (3), Autosomal recessive
<i>C12orf57</i>	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<i>C19orf12</i>	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
<i>C1QA</i>	120550	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QB</i>	120570	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QC</i>	120575	C1q deficiency, 613652 (3), Autosomal recessive
<i>C2CD3</i>	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
<i>C3</i>	120700	C3 deficiency, 613779 (3), Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 (3), Autosomal dominant; {Macular degeneration, age-related, 9}, 611378 (3)
<i>C5</i>	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant
<i>C6</i>	217050	C6 deficiency, 612446 (3), Autosomal recessive; Combined C6/C7 deficiency (3)
<i>C7</i>	217070	C7 deficiency, 610102 (3)
<i>C8A</i>	120950	C8 deficiency, type I, 613790 (3), Autosomal recessive
<i>C8B</i>	120960	C8 deficiency, type II, 613789 (3), Autosomal recessive
<i>CA2</i>	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 (3), Autosomal recessive
<i>CABP4</i>	608965	Cone-rod synaptic disorder, congenital nonprogressive, 610427 (3), Autosomal recessive
<i>CACNA1F</i>	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
<i>CANT1</i>	613165	Desbuquois dysplasia 1, 251450 (3), Autosomal recessive; Epiphyseal dysplasia, multiple, 7, 617719 (3), Autosomal recessive

<i>CAPN3</i>	114240	Muscular dystrophy, limb-girdle, autosomal recessive 1, 253600 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal dominant 4, 618129 (3), Autosomal dominant
<i>CARD11</i>	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
<i>CARD9</i>	607212	Immunodeficiency 103, susceptibility to fungal infection, 212050 (3), Autosomal recessive
<i>CARS2</i>	612800	Combined oxidative phosphorylation deficiency 27, 616672 (3), Autosomal recessive
<i>CASK</i>	300172	Intellectual developmental disorder, with or without nystagmus, 300422 (3); Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked dominant; FG syndrome 4, 300422 (3)
<i>CASQ2</i>	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938 (3), Autosomal recessive
<i>CASR</i>	601199	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198 (3), Autosomal dominant; {Epilepsy idiopathic generalized, susceptibility to, 8}, 612899 (3); Hyperparathyroidism, neonatal, 239200 (3), Autosomal dominant, Autosomal recessive; Hypocalcemia, autosomal dominant, 601198 (3), Autosomal dominant; Hypocalciuric hypercalcemia, type I, 145980 (3), Autosomal dominant
<i>CAV3</i>	601253	Myopathy, distal, Tateyama type, 614321 (3), Autosomal dominant; Creatine phosphokinase, elevated serum, 123320 (3), Autosomal dominant; Cardiomyopathy, familial hypertrophic, 192600 (3), Digenic dominant, Autosomal dominant; Rippling muscle disease 2, 606072 (3), Autosomal dominant; Long QT syndrome 9, 611818 (3), Autosomal dominant
<i>CAVIN1</i>	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
<i>CBS</i>	613381	Thrombosis, hyperhomocysteinemic, 236200 (3), Autosomal recessive; Homocystinuria, B6-responsive and nonresponsive types, 236200 (3), Autosomal recessive
<i>CC2D1A</i>	610055	Intellectual developmental disorder, autosomal recessive 3, 608443 (3), Autosomal recessive
<i>CC2D2A</i>	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
<i>CCBE1</i>	612753	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 (3), Autosomal recessive
<i>CCDC103</i>	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
<i>CCDC115</i>	613734	Congenital disorder of glycosylation, type IIo, 616828 (3), Autosomal recessive

<i>CCDC28B</i>	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Digenic recessive, Autosomal recessive
<i>CCDC39</i>	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
<i>CCDC40</i>	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
<i>CCDC8</i>	614145	3-M syndrome 3, 614205 (3), Autosomal recessive
<i>CCDC88C</i>	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
<i>CCN6</i>	603400	Progressive pseudorheumatoid dysplasia, 208230 (3), Autosomal recessive
<i>CCNO</i>	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
<i>CCNQ</i>	300708	STAR syndrome, 300707 (3), X-linked dominant
<i>CD19</i>	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
<i>CD27</i>	186711	Lymphoproliferative syndrome 2, 615122 (3), Autosomal recessive
<i>CD3D</i>	186790	Immunodeficiency 19, severe combined, 615617 (3), Autosomal recessive
<i>CD3E</i>	186830	Immunodeficiency 18, 615615 (3), Autosomal recessive; Immunodeficiency 18, SCID variant, 615615 (3), Autosomal recessive
<i>CD3G</i>	186740	Immunodeficiency 17, CD3 gamma deficient, 615607 (3), Autosomal recessive
<i>CD40</i>	109535	Immunodeficiency with hyper-IgM, type 3, 606843 (3), Autosomal recessive
<i>CD40LG</i>	300386	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3), X-linked recessive
<i>CD55</i>	125240	[Blood group Cromer], 613793 (3), Autosomal recessive; Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300 (3), Autosomal recessive
<i>CD59</i>	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300 (3), Autosomal recessive
<i>CD81</i>	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
<i>CD96</i>	606037	C syndrome, 211750 (3), Autosomal dominant
<i>CDAN1</i>	607465	Dyserythropoietic anemia, congenital, type Ia, 224120 (3), Autosomal recessive
<i>CDC45</i>	603465	Meier-Gorlin syndrome 7, 617063 (3), Autosomal recessive
<i>CDC6</i>	602627	?Meier-Gorlin syndrome 5, 613805 (3), Autosomal recessive
<i>CDH11</i>	600023	Teebi hypertelorism syndrome 2, 619736 (3), Autosomal dominant; Elshahy-Waters syndrome, 211380 (3), Autosomal recessive
<i>CDH23</i>	605516	Usher syndrome, type 1D, 601067 (3), Digenic recessive, Autosomal recessive; {Pituitary adenoma 5, multiple types}, 617540 (3), Autosomal dominant; Usher syndrome, type 1D/F digenic, 601067 (3), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 12, 601386 (3), Autosomal recessive

<i>CDH3</i>	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
<i>CDK10</i>	603464	Al Kaissi syndrome, 617694 (3), Autosomal recessive
<i>CDK5RAP2</i>	608201	Microcephaly 3, primary, autosomal recessive, 604804 (3), Autosomal recessive
<i>CDKL5</i>	300203	Developmental and epileptic encephalopathy 2, 300672 (3), X-linked dominant
<i>CDT1</i>	605525	Meier-Gorlin syndrome 4, 613804 (3), Autosomal recessive
<i>CENPJ</i>	609279	Microcephaly 6, primary, autosomal recessive, 608393 (3), Autosomal recessive; ?Seckel syndrome 4, 613676 (3), Autosomal recessive
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
<i>CEP120</i>	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
<i>CEP152</i>	613529	Microcephaly 9, primary, autosomal recessive, 614852 (3), Autosomal recessive; Seckel syndrome 5, 613823 (3), Autosomal recessive
<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP290</i>	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP57</i>	607951	Mosaic variegated aneuploidy syndrome 2, 614114 (3), Autosomal recessive
<i>CEP78</i>	617110	Cone-rod dystrophy and hearing loss, 617236 (3), Autosomal recessive
<i>CERKL</i>	608381	Retinitis pigmentosa 26, 608380 (3), Autosomal recessive
<i>CERS3</i>	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
<i>CFAP410</i>	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
<i>CFAP418</i>	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive
<i>CFD</i>	134350	Complement factor D deficiency, 613912 (3), Autosomal recessive
<i>CFH</i>	134370	{Macular degeneration, age-related, 4}, 610698 (3); Basal laminar drusen, 126700 (3), Autosomal dominant; Complement factor H deficiency, 609814 (3), Autosomal dominant, Autosomal recessive; {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 (3), Autosomal dominant, Autosomal recessive

<i>CFI</i>	217030	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 (3), Autosomal dominant; {Macular degeneration, age-related, 13, susceptibility to}, 615439 (3), Autosomal dominant; Complement factor I deficiency, 610984 (3), Autosomal recessive
<i>CFL2</i>	601443	Nemaline myopathy 7, autosomal recessive, 610687 (3), Autosomal recessive
<i>CFP</i>	300383	Properdin deficiency, X-linked, 312060 (3), X-linked recessive
<i>CFTR</i>	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
<i>CHAT</i>	118490	Myasthenic syndrome, congenital, 6, presynaptic, 254210 (3), Autosomal recessive
<i>CHKB</i>	612395	Muscular dystrophy, congenital, megaconial type, 602541 (3), Autosomal recessive
<i>CHM</i>	300390	Choroideremia, 303100 (3), X-linked
<i>CHMP1A</i>	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
<i>CHRNA1</i>	100690	Myasthenic syndrome, congenital, 1B, fast-channel, 608930 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 1A, slow-channel, 601462 (3), Autosomal dominant; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive
<i>CHRNB1</i>	100710	?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314 (3), Autosomal recessive; Myasthenic syndrome, congenital, 2A, slow-channel, 616313 (3), Autosomal dominant
<i>CHRND</i>	100720	?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323 (3), Autosomal recessive; Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Myasthenic syndrome, congenital, 3B, fast-channel, 616322 (3), Autosomal recessive; ?Myasthenic syndrome, congenital, 3A, slow-channel, 616321 (3), Autosomal dominant
<i>CHRNE</i>	100725	Myasthenic syndrome, congenital, 4A, slow-channel, 605809 (3), Autosomal dominant, Autosomal recessive; Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931 (3), Autosomal recessive; Myasthenic syndrome, congenital, 4B, fast-channel, 616324 (3), Autosomal recessive
<i>CHRNG</i>	100730	Multiple pterygium syndrome, lethal type, 253290 (3), Autosomal recessive; Escobar syndrome, 265000 (3), Autosomal recessive
<i>CHST14</i>	608429	Ehlers-Danlos syndrome, musculocontractural type 1, 601776 (3), Autosomal recessive
<i>CHST3</i>	603799	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095 (3), Autosomal recessive

<i>CHST6</i>	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
<i>CHSY1</i>	608183	Temtamy preaxial brachydactyly syndrome, 605282 (3), Autosomal recessive
<i>CIB2</i>	605564	Deafness, autosomal recessive 48, 609439 (3), Autosomal recessive; Usher syndrome, type II, 614869 (3), Autosomal recessive
<i>CIITA</i>	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive
<i>CILK1</i>	612325	{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
<i>CISD2</i>	611507	Wolfram syndrome 2, 604928 (3), Autosomal recessive
<i>CIT</i>	605629	Microcephaly 17, primary, autosomal recessive, 617090 (3), Autosomal recessive
<i>CKAP2L</i>	616174	Filippi syndrome, 272440 (3), Autosomal recessive
<i>CLCF1</i>	607672	Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive
<i>CLCN1</i>	118425	Myotonia levior, recessive (3); Myotonia congenita, recessive, 255700 (3), Autosomal recessive; Myotonia congenita, dominant, 160800 (3), Autosomal dominant
<i>CLCN2</i>	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
<i>CLCN4</i>	302910	Raynaud-Claes syndrome, 300114 (3), X-linked dominant
<i>CLCN5</i>	300008	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990 (3), X-linked recessive; Hypophosphatemic rickets, 300554 (3), X-linked recessive; Dent disease 1, 300009 (3), X-linked recessive; Nephrolithiasis, type I, 310468 (3), X-linked recessive
<i>CLCN7</i>	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
<i>CLCNKB</i>	602023	Bartter syndrome, type 3, 607364 (3), Autosomal recessive; Bartter syndrome, type 4b, digenic, 613090 (3), Digenic recessive
<i>CLDN1</i>	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
<i>CLDN10</i>	617579	HELIX syndrome, 617671 (3), Autosomal recessive
<i>CLDN14</i>	605608	Deafness, autosomal recessive 29, 614035 (3), Autosomal recessive
<i>CLDN16</i>	603959	Hypomagnesemia 3, renal, 248250 (3), Autosomal recessive
<i>CLDN19</i>	610036	Hypomagnesemia 5, renal, with ocular involvement, 248190 (3), Autosomal recessive
<i>CLMP</i>	611693	Congenital short bowel syndrome, 615237 (3), Autosomal recessive

<i>CLN3</i>	607042	Ceroid lipofuscinosis, neuronal, 3, 204200 (3), Autosomal recessive
<i>CLN5</i>	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
<i>CLN6</i>	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
<i>CLN8</i>	607837	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 8, 600143 (3), Autosomal recessive
<i>CLP1</i>	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
<i>CLPB</i>	616254	Neutropenia, severe congenital, 9, autosomal dominant, 619813 (3), Autosomal dominant; 3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271 (3), Autosomal recessive; 3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 (3), Autosomal dominant
<i>CLPP</i>	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
<i>CLRN1</i>	606397	Usher syndrome, type 3A, 276902 (3), Autosomal recessive; Retinitis pigmentosa 61, 614180 (3)
<i>CNGA1</i>	123825	Retinitis pigmentosa 49, 613756 (3)
<i>CNGA3</i>	600053	Achromatopsia 2, 216900 (3), Autosomal recessive
<i>CNGB1</i>	600724	Retinitis pigmentosa 45, 613767 (3), Autosomal recessive
<i>CNGB3</i>	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
<i>CNNM4</i>	607805	Jalili syndrome, 217080 (3), Autosomal recessive
<i>CNTN1</i>	600016	?Myopathy, congenital, Compton-North, 612540 (3), Autosomal recessive
<i>CNTNAP1</i>	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<i>CNTNAP2</i>	604569	Pitt-Hopkins like syndrome 1, 610042 (3), Autosomal recessive; {Autism susceptibility 15}, 612100 (3)
<i>COA8</i>	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
<i>COG1</i>	606973	Congenital disorder of glycosylation, type IIg, 611209 (3), Autosomal recessive
<i>COG4</i>	606976	Congenital disorder of glycosylation, type IIj, 613489 (3), Autosomal recessive; Saul-Wilson syndrome, 618150 (3), Autosomal dominant
<i>COG6</i>	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
<i>COG7</i>	606978	Congenital disorder of glycosylation, type IIe, 608779 (3), Autosomal recessive
<i>COG8</i>	606979	Congenital disorder of glycosylation, type IIh, 611182 (3)
<i>COL11A1</i>	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3)

<i>COL11A2</i>	120290	Deafness, autosomal dominant 13, 601868 (3), Autosomal dominant; Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 (3), Autosomal recessive; Fibrochondrogenesis 2, 614524 (3), Autosomal dominant, Autosomal recessive; Deafness, autosomal recessive 53, 609706 (3), Autosomal recessive; Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 (3), Autosomal dominant
<i>COL17A1</i>	113811	Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
<i>COL1A2</i>	120160	Osteogenesis imperfecta, type III, 259420 (3), Autosomal dominant; {Osteoporosis, postmenopausal}, 166710 (3), Autosomal dominant; Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 (3), Autosomal dominant; Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120 (3), Autosomal dominant; Ehlers-Danlos syndrome, cardiac valvular type, 225320 (3), Autosomal recessive; Osteogenesis imperfecta, type IV, 166220 (3), Autosomal dominant; Osteogenesis imperfecta, type II, 166210 (3), Autosomal dominant
<i>COL27A1</i>	608461	Steel syndrome, 615155 (3), Autosomal recessive
<i>COL2A1</i>	120140	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant
<i>COL4A3</i>	120070	Hematuria, benign familial, 141200 (3), Autosomal dominant; Alport syndrome 3, autosomal dominant, 104200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive
<i>COL4A4</i>	120131	Hematuria, familial benign, 141200 (3), Autosomal dominant; Alport syndrome 2, autosomal recessive, 203780 (3), Autosomal recessive
<i>COL4A5</i>	303630	Alport syndrome 1, X-linked, 301050 (3), X-linked dominant

<i>COL6A1</i>	120220	Bethlem myopathy 1, 158810 (3), Autosomal dominant, Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal dominant, Autosomal recessive
<i>COL6A2</i>	120240	Bethlem myopathy 1, 158810 (3), Autosomal dominant, Autosomal recessive; ?Myosclerosis, congenital, 255600 (3), Autosomal recessive; Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal dominant, Autosomal recessive
<i>COL6A3</i>	120250	Ullrich congenital muscular dystrophy 1, 254090 (3), Autosomal dominant, Autosomal recessive; Dystonia 27, 616411 (3), Autosomal recessive; Bethlem myopathy 1, 158810 (3), Autosomal dominant, Autosomal recessive
<i>COL7A1</i>	120120	EBD, localisata variant (3); Epidermolysis bullosa, pretibial, 131850 (3), Autosomal dominant, Autosomal recessive; Transient bullous of the newborn, 131705 (3), Autosomal dominant, Autosomal recessive; EBD, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal dominant, Autosomal recessive; EBD inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant
<i>COL9A2</i>	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
<i>COLEC11</i>	612502	3MC syndrome 2, 265050 (3), Autosomal recessive
<i>COLQ</i>	603033	Myasthenic syndrome, congenital, 5, 603034 (3), Autosomal recessive
<i>COQ2</i>	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
<i>COQ4</i>	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
<i>COQ6</i>	614647	Coenzyme Q10 deficiency, primary, 6, 614650 (3), Autosomal recessive
<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ8B</i>	615567	Nephrotic syndrome, type 9, 615573 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>COX10</i>	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
<i>COX15</i>	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
<i>COX20</i>	614698	Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive
<i>COX6B1</i>	124089	Mitochondrial complex IV deficiency, nuclear type 7, 619051 (3), Autosomal recessive
<i>COX7B</i>	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant

<i>CP</i>	117700	Cerebellar ataxia, 604290 (3), Autosomal recessive; [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive
<i>CPAMD8</i>	608841	Anterior segment dysgenesis 8, 617319 (3), Autosomal recessive
<i>CPLANE1</i>	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
<i>CPS1</i>	608307	Carbamoylphosphate synthetase I deficiency, 237300 (3), Autosomal recessive; {Pulmonary hypertension, neonatal, susceptibility to}, 615371 (3)
<i>CPT1A</i>	600528	CPT deficiency, hepatic, type IA, 255120 (3), Autosomal recessive
<i>CPT2</i>	600650	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212 (3), Autosomal dominant, Autosomal recessive; CPT II deficiency, infantile, 600649 (3), Autosomal recessive; CPT II deficiency, lethal neonatal, 608836 (3), Autosomal recessive; CPT II deficiency, myopathic, stress-induced, 255110 (3), Autosomal dominant, Autosomal recessive
<i>CRB1</i>	604210	Leber congenital amaurosis 8, 613835 (3), Autosomal recessive; Retinitis pigmentosa-12, 600105 (3), Autosomal recessive; Pigmented paravenous chorioretinal atrophy, 172870 (3), Autosomal dominant
<i>CRB2</i>	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
<i>CRLF1</i>	604237	Cold-induced sweating syndrome 1, 272430 (3), Autosomal recessive
<i>CRPPA</i>	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
<i>CRTAP</i>	605497	Osteogenesis imperfecta, type VII, 610682 (3), Autosomal recessive
<i>CRYAB</i>	123590	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>CSTB</i>	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<i>CTC1</i>	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
<i>CTH</i>	607657	Cystathioninuria, 219500 (3), Autosomal recessive
<i>CTNS</i>	606272	Cystinosis, nephropathic, 219800 (3), Autosomal recessive; Cystinosis, ocular nonnephropathic, 219750 (3), Autosomal recessive; Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3), Autosomal recessive; Cystinosis, atypical nephropathic, 219800 (3), Autosomal recessive
<i>CTPS1</i>	123860	Immunodeficiency 24, 615897 (3), Autosomal recessive

<i>CTSA</i>	613111	Galactosialidosis, 256540 (3), Autosomal recessive
<i>CTSC</i>	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
<i>CTSD</i>	116840	Ceroid lipofuscinosis, neuronal, 10, 610127 (3), Autosomal recessive
<i>CTSF</i>	603539	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362 (3), Autosomal recessive
<i>CTSK</i>	601105	Pycnodysostosis, 265800 (3), Autosomal recessive
<i>CUBN</i>	602997	[Proteinuria, chronic benign], 618884 (3), Autosomal recessive; Imerslund-Grasbeck syndrome 1, 261100 (3), Autosomal recessive
<i>CUL4B</i>	300304	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354 (3), X-linked recessive
<i>CUL7</i>	609577	3-M syndrome 1, 273750 (3), Autosomal recessive
<i>CWC27</i>	617170	Retinitis pigmentosa with or without skeletal anomalies, 250410 (3), Autosomal recessive
<i>CYB5R3</i>	613213	Methemoglobinemia, type I, 250800 (3), Autosomal recessive; Methemoglobinemia, type II, 250800 (3), Autosomal recessive
<i>CYBA</i>	608508	Chronic granulomatous disease 4, autosomal recessive, 233690 (3), Autosomal recessive
<i>CYBB</i>	300481	Immunodeficiency 34, mycobacteriosis, X-linked, 300645 (3), X-linked recessive; Chronic granulomatous disease, X-linked, 306400 (3), X-linked recessive
<i>CYP11A1</i>	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
<i>CYP11B1</i>	610613	Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive
<i>CYP11B2</i>	124080	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive
<i>CYP17A1</i>	609300	17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive
<i>CYP19A1</i>	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
<i>CYP1B1</i>	601771	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300 (3), Autosomal recessive; Anterior segment dysgenesis 6, multiple subtypes, 617315 (3), Autosomal recessive
<i>CYP21A2</i>	613815	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
<i>CYP24A1</i>	126065	Hypercalcemia, infantile, 1, 143880 (3), Autosomal recessive
<i>CYP26C1</i>	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive

<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP27B1</i>	609506	Vitamin D-dependent rickets, type I, 264700 (3), Autosomal recessive
<i>CYP2U1</i>	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<i>CYP4F22</i>	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
<i>CYP4V2</i>	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
<i>CYP7B1</i>	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
<i>D2HGDH</i>	609186	D-2-hydroxyglutaric aciduria, 600721 (3), Autosomal recessive
<i>DARS1</i>	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DBH</i>	609312	Orthostatic hypotension 1, due to DBH deficiency, 223360 (3), Autosomal recessive
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DCDC2</i>	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
<i>DCHS1</i>	603057	Mitral valve prolapse 2, 607829 (3), Autosomal dominant; Van Maldergem syndrome 1, 601390 (3), Autosomal recessive
<i>DCLRE1C</i>	605988	Severe combined immunodeficiency, Athabaskan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
<i>DCX</i>	300121	Subcortical laminar heterotopia, X-linked, 300067 (3), X-linked; Lissencephaly, X-linked, 300067 (3), X-linked
<i>DDB2</i>	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
<i>DDC</i>	107930	Aromatic L-amino acid decarboxylase deficiency, 608643 (3), Autosomal recessive
<i>DDHD2</i>	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<i>DDR2</i>	191311	Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive
<i>DDX11</i>	601150	Warsaw breakage syndrome, 613398 (3), Autosomal recessive
<i>DDX59</i>	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
<i>DENND5A</i>	617278	Developmental and epileptic encephalopathy 49, 617281 (3), Autosomal recessive

<i>DES</i>	125660	Scapulooperoneal syndrome, neurogenic, Kaeser type, 181400 (3), Autosomal dominant; Cardiomyopathy, dilated, 1I, 604765 (3), Autosomal dominant; Myopathy, myofibrillar, 1, 601419 (3), Autosomal dominant, Autosomal recessive
<i>DGAT1</i>	604900	?Diarrhea 7, protein-losing enteropathy type, 615863 (3), Autosomal recessive
<i>DGKE</i>	601440	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 (3), Autosomal recessive; Nephrotic syndrome, type 7, 615008 (3), Autosomal recessive
<i>DGUOK</i>	601465	Portal hypertension, noncirrhotic, 1, 617068 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 (3), Autosomal recessive
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
<i>DHDDS</i>	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
<i>DHH</i>	605423	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 (3), Autosomal recessive; 46XY sex reversal 7, 233420 (3), Autosomal recessive
<i>DHODH</i>	126064	Miller syndrome, 263750 (3), Autosomal recessive
<i>DIS3L2</i>	614184	Perlman syndrome, 267000 (3), Autosomal recessive
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DLG3</i>	300189	Intellectual developmental disorder, X-linked 90, 300850 (3), X-linked recessive
<i>DLL3</i>	602768	Spondylocostal dysostosis 1, autosomal recessive, 277300 (3), Autosomal recessive
<i>DLX5</i>	600028	Split-hand/foot malformation 1, 183600 (3), Autosomal dominant; ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 (3), Autosomal recessive
<i>DMD</i>	300377	Becker muscular dystrophy, 300376 (3), X-linked recessive; Cardiomyopathy, dilated, 3B, 302045 (3), X-linked; Duchenne muscular dystrophy, 310200 (3), X-linked recessive
<i>DMP1</i>	600980	Hypophosphatemic rickets, AR, 241520 (3), Autosomal recessive
<i>DNAAF1</i>	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
<i>DNAAF11</i>	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
<i>DNAAF3</i>	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
<i>DNAAF4</i>	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive

<i>DNAAF5</i>	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
<i>DNAAF6</i>	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
<i>DNAH11</i>	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
<i>DNAH5</i>	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
<i>DNAH9</i>	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
<i>DNAI1</i>	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
<i>DNAI2</i>	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
<i>DNAJC12</i>	606060	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384 (3), Autosomal recessive
<i>DNAJC19</i>	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<i>DNAJC21</i>	617048	Bone marrow failure syndrome 3, 617052 (3), Autosomal recessive
<i>DNAJC6</i>	608375	Parkinson disease 19a, juvenile-onset, 615528 (3), Autosomal recessive; Parkinson disease 19b, early-onset, 615528 (3), Autosomal recessive
<i>DNAL1</i>	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
<i>DNM2</i>	602378	Centronuclear myopathy 1, 160150 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal type 2M, 606482 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3), Autosomal dominant; Lethal congenital contracture syndrome 5, 615368 (3), Autosomal recessive
<i>DNMT3B</i>	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860 (3), Autosomal recessive; Facioscapulohumeral muscular dystrophy 4, digenic, 619478 (3), Digenic dominant
<i>DOCK2</i>	603122	Immunodeficiency 40, 616433 (3), Autosomal recessive
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DOCK8</i>	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
<i>DOK7</i>	610285	Fetal akinesia deformation sequence 3, 618389 (3), Autosomal recessive; Myasthenic syndrome, congenital, 10, 254300 (3), Autosomal recessive
<i>DOLK</i>	610746	Congenital disorder of glycosylation, type Im, 610768 (3), Autosomal recessive
<i>DONSON</i>	611428	Microcephaly, short stature, and limb abnormalities, 617604 (3), Autosomal recessive; Microcephaly-micromelia syndrome, 251230 (3), Autosomal recessive
<i>DPAGT1</i>	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750 (3), Autosomal recessive; Congenital disorder of glycosylation, type Ij, 608093 (3), Autosomal recessive
<i>DPH1</i>	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive

<i>DPM1</i>	603503	Congenital disorder of glycosylation, type 1e, 608799 (3), Autosomal recessive
<i>DPYD</i>	612779	Dihydropyrimidine dehydrogenase deficiency, 274270 (3), Autosomal recessive; 5-fluorouracil toxicity, 274270 (3), Autosomal recessive
<i>DSC2</i>	125645	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal dominant, Autosomal recessive; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal dominant, Autosomal recessive
<i>DSG1</i>	125670	Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive
<i>DSP</i>	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive
<i>DSTYK</i>	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
<i>DUOX2</i>	606759	Thyroid dysmorphogenesis 6, 607200 (3), Autosomal recessive
<i>DUOXA2</i>	612772	Thyroid dysmorphogenesis 5, 274900 (3), Autosomal recessive
<i>DYM</i>	607461	Smith-McCort dysplasia, 607326 (3), Autosomal recessive; Dyggve-Melchior-Clausen disease, 223800 (3), Autosomal recessive
<i>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Digenic recessive, Autosomal recessive
<i>DYNC2I1</i>	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
<i>DYNC2I2</i>	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
<i>DYNC2LI1</i>	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
<i>DYSF</i>	603009	Muscular dystrophy, limb-girdle, autosomal recessive 2, 253601 (3), Autosomal recessive; Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive
<i>EARS2</i>	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<i>EBP</i>	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant
<i>ECEL1</i>	605896	Arthrogryposis, distal, type 5D, 615065 (3), Autosomal recessive
<i>ECHS1</i>	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive

<i>ECM1</i>	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
<i>EDA</i>	300451	Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive
<i>EDAR</i>	604095	[Hair morphology 1, hair thickness], 612630 (3); Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive
<i>EDARADD</i>	606603	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant
<i>EDN3</i>	131242	Waardenburg syndrome, type 4B, 613265 (3), Autosomal dominant, Autosomal recessive; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant
<i>EDNRB</i>	131244	{Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ?ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal dominant, Autosomal recessive
<i>EFEMP2</i>	604633	Cutis laxa, autosomal recessive, type 1B, 614437 (3), Autosomal recessive
<i>EFNB1</i>	300035	Craniofrontonasal dysplasia, 304110 (3), X-linked dominant
<i>EGR2</i>	129010	Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 1D, 607678 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 1, 605253 (3), Autosomal dominant, Autosomal recessive
<i>EIF2AK3</i>	604032	Wolcott-Rallison syndrome, 226980 (3), Autosomal recessive
<i>EIF2AK4</i>	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
<i>EIF2B1</i>	606686	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B2</i>	606454	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive; Ovarioleukodystrophy, 603896 (3), Autosomal recessive
<i>EIF2B3</i>	606273	Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B4</i>	606687	Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2B5</i>	603945	Ovarioleukodystrophy, 603896 (3), Autosomal recessive; Leukoencephalopathy with vanishing white matter, 603896 (3), Autosomal recessive
<i>EIF2S3</i>	300161	MEHMO syndrome, 300148 (3), X-linked recessive
<i>EIF4A3</i>	608546	Robin sequence with cleft mandible and limb anomalies, 268305 (3), Autosomal recessive

<i>ELAC2</i>	605367	{Prostate cancer, hereditary, 2, susceptibility to}, 614731 (3); Combined oxidative phosphorylation deficiency 17, 615440 (3), Autosomal recessive
<i>ELOVL4</i>	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive
<i>ELP1</i>	603722	Dysautonomia, familial, 223900 (3), Autosomal recessive; Medulloblastoma, 155255 (3), Autosomal dominant, Somatic mutation, Autosomal recessive
<i>ELP2</i>	616054	Intellectual developmental disorder, autosomal recessive 58, 617270 (3), Autosomal recessive
<i>EMD</i>	300384	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300 (3), X-linked recessive
<i>EMG1</i>	611531	Bowen-Conradi syndrome, 211180 (3), Autosomal recessive
<i>EML1</i>	602033	Band heterotopia, 600348 (3), Autosomal recessive
<i>ENPP1</i>	173335	{Obesity, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
<i>ENTPD1</i>	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<i>EOGT</i>	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
<i>EPB42</i>	177070	Spherocytosis, type 5, 612690 (3)
<i>EPCAM</i>	185535	Colorectal cancer, hereditary nonpolyposis, type 8, 613244 (3), Autosomal dominant; Diarrhea 5, with tufting enteropathy, congenital, 613217 (3), Autosomal recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>EPM2A</i>	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
<i>ERBB3</i>	190151	?Lethal congenital contractural syndrome 2, 607598 (3), Autosomal recessive; {?Erythroleukemia, familial, susceptibility to}, 133180 (3), Autosomal dominant; Visceral neuropathy, familial, 1, autosomal recessive, 243180 (3), Autosomal recessive
<i>ERCC1</i>	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
<i>ERCC2</i>	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
<i>ERCC3</i>	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive

<i>ERCC4</i>	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
<i>ERCC5</i>	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
<i>ERCC6</i>	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
<i>ERCC6L2</i>	615667	Bone marrow failure syndrome 2, 615715 (3), Autosomal recessive
<i>ERCC8</i>	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
<i>ERLIN1</i>	611604	Spastic paraplegia 62, 615681 (3), Autosomal recessive
<i>ERLIN2</i>	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
<i>ESCO2</i>	609353	Juberg-Hayward syndrome, 216100 (3), Autosomal recessive; Roberts-SC phocomelia syndrome, 268300 (3), Autosomal recessive
<i>ESRRB</i>	602167	Deafness, autosomal recessive 35, 608565 (3), Autosomal recessive
<i>ETFA</i>	608053	Glutaric acidemia IIA, 231680 (3), Autosomal recessive
<i>ETFB</i>	130410	Glutaric acidemia IIB, 231680 (3), Autosomal recessive
<i>ETFDH</i>	231675	Glutaric acidemia IIC, 231680 (3), Autosomal recessive
<i>ETHE1</i>	608451	Ethylmalonic encephalopathy, 602473 (3), Autosomal recessive
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>EXOSC8</i>	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
<i>EXTL3</i>	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
<i>EYS</i>	612424	Retinitis pigmentosa 25, 602772 (3), Autosomal recessive
<i>F11</i>	264900	Factor XI deficiency, autosomal dominant, 612416 (3); Factor XI deficiency, autosomal recessive, 612416 (3)
<i>F13A1</i>	134570	Factor XIII A deficiency, 613225 (3), Autosomal recessive; {Myocardial infarction, protection against}, 608446 (3); {Venous thrombosis, protection against}, 188050 (3), Autosomal dominant

<i>F2</i>	176930	Hypoprothrombinemia, 613679 (3), Autosomal recessive; {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 (3), Autosomal dominant; Dysprothrombinemia, 613679 (3), Autosomal recessive; Thrombophilia 1 due to thrombin defect, 188050 (3), Autosomal dominant; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial
<i>F5</i>	612309	Thrombophilia 2 due to activated protein C resistance, 188055 (3), Autosomal dominant; {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 (3), Autosomal dominant; {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055 (3), Autosomal dominant; {Budd-Chiari syndrome}, 600880 (3), Autosomal recessive; {Stroke, ischemic, susceptibility to}, 601367 (3), Multifactorial; Factor V deficiency, 227400 (3), Autosomal recessive
<i>F7</i>	613878	{Myocardial infarction, decreased susceptibility to}, 608446 (3); Factor VII deficiency, 227500 (3), Autosomal recessive
<i>F8</i>	300841	Thrombophilia 13, X-linked, due to factor VIII defect, 301071 (3); Hemophilia A, 306700 (3), X-linked recessive
<i>F9</i>	300746	{Deep venous thrombosis, protection against}, 300807 (3), X-linked recessive; Hemophilia B, 306900 (3), X-linked recessive; Thrombophilia 8, X-linked, due to factor IX defect, 300807 (3), X-linked recessive; {Warfarin sensitivity}, 301052 (3), X-linked
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FAH</i>	613871	Tyrosinemia, type I, 276700 (3), Autosomal recessive
<i>FAM161A</i>	613596	Retinitis pigmentosa 28, 606068 (3)
<i>FAM20C</i>	611061	Raine syndrome, 259775 (3), Autosomal recessive
<i>FANCA</i>	607139	Fanconi anemia, complementation group A, 227650 (3), Autosomal recessive
<i>FANCB</i>	300515	Fanconi anemia, complementation group B, 300514 (3), X-linked recessive
<i>FANCC</i>	613899	Fanconi anemia, complementation group C, 227645 (3), Autosomal recessive
<i>FANCD2</i>	613984	Fanconi anemia, complementation group D2, 227646 (3), Autosomal recessive
<i>FANCE</i>	613976	Fanconi anemia, complementation group E, 600901 (3), Autosomal recessive
<i>FANCF</i>	613897	Fanconi anemia, complementation group F, 603467 (3), Autosomal recessive
<i>FANCG</i>	602956	Fanconi anemia, complementation group G, 614082 (3), Autosomal recessive
<i>FANCI</i>	611360	Fanconi anemia, complementation group I, 609053 (3), Autosomal recessive
<i>FANCL</i>	608111	Fanconi anemia, complementation group L, 614083 (3), Autosomal recessive

<i>FARS2</i>	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
<i>FAS</i>	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant
<i>FASTKD2</i>	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
<i>FAT4</i>	612411	Van Maldergem syndrome 2, 615546 (3), Autosomal recessive; Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 (3), Autosomal recessive
<i>FBLN5</i>	604580	Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant
<i>FBP1</i>	611570	Fructose-1,6-bisphosphatase deficiency, 229700 (3), Autosomal recessive
<i>FBXL4</i>	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<i>FBXO7</i>	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
<i>FECH</i>	612386	Protoporphyrinemia, erythropoietic, 1, 177000 (3), Autosomal recessive
<i>FERMT1</i>	607900	Kindler syndrome, 173650 (3), Autosomal recessive
<i>FERMT3</i>	607901	Leukocyte adhesion deficiency, type III, 612840 (3), Autosomal recessive
<i>FGA</i>	134820	Hypodysfibrinogenemia, congenital, 616004 (3); Dysfibrinogenemia, congenital, 616004 (3); Amyloidosis, familial visceral, 105200 (3), Autosomal dominant; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
<i>FGB</i>	134830	Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Dysfibrinogenemia, congenital, 616004 (3); Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
<i>FGD1</i>	300546	Intellectual developmental disorder, X-linked syndromic 16, 305400 (3), X-linked recessive; Aarskog-Scott syndrome, 305400 (3), X-linked recessive
<i>FGD4</i>	611104	Charcot-Marie-Tooth disease, type 4H, 609311 (3), Autosomal recessive
<i>FGF3</i>	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3), Autosomal recessive

<i>FGFR2</i>	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3); Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chatzen syndrome, 101400 (3), Autosomal dominant
<i>FGFR3</i>	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal dominant, Autosomal recessive; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
<i>FGG</i>	134850	Dysfibrinogenemia, congenital, 616004 (3); Hypodysfibrinogenemia, 616004 (3); Hypofibrinogenemia, congenital, 202400 (3), Autosomal recessive; Afibrinogenemia, congenital, 202400 (3), Autosomal recessive
<i>FH</i>	136850	Leiomyomatosis and renal cell cancer, 150800 (3), Autosomal dominant; Fumarase deficiency, 606812 (3), Autosomal recessive
<i>FHL1</i>	300163	Myopathy, X-linked, with postural muscle atrophy, 300696 (3), X-linked recessive; Emery-Dreifuss muscular dystrophy 6, X-linked, 300696 (3), X-linked recessive; ?Uruguay faciocardiomusculoskeletal syndrome, 300280 (3), X-linked recessive; Scapulooperoneal myopathy, X-linked dominant, 300695 (3), X-linked dominant; Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718 (3), X-linked; Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717 (3), X-linked dominant
<i>FIG4</i>	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
<i>FKBP10</i>	607063	Osteogenesis imperfecta, type XI, 610968 (3), Autosomal recessive; Bruck syndrome 1, 259450 (3), Autosomal recessive

<i>FKBP14</i>	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive
<i>FKRP</i>	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
<i>FKTN</i>	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive
<i>FLAD1</i>	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100 (3), Autosomal recessive
<i>FLNA</i>	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
<i>FLNB</i>	603381	Larsen syndrome, 150250 (3), Autosomal dominant; Atelosteogenesis, type I, 108720 (3), Autosomal dominant; Atelosteogenesis, type III, 108721 (3), Autosomal dominant; Spondylocarpotarsal synostosis syndrome, 272460 (3), Autosomal recessive; Boomerang dysplasia, 112310 (3), Autosomal dominant
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FLVCR2</i>	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790 (3), Autosomal recessive
<i>FMO3</i>	136132	Trimethylaminuria, 602079 (3), Autosomal recessive
<i>FMR1</i>	309550	Fragile X tremor/ataxia syndrome, 300623 (3), X-linked dominant; Fragile X syndrome, 300624 (3), X-linked dominant; Premature ovarian failure 1, 311360 (3), X-linked
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FOXE1</i>	602617	Bamforth-Lazarus syndrome, 241850 (3), Autosomal recessive; {Thyroid cancer, nonmedullary, 4}, 616534 (3), Autosomal dominant

<i>FOXE3</i>	601094	Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3)
<i>FOXN1</i>	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3), Autosomal dominant; T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
<i>FOXP3</i>	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
<i>FOXRED1</i>	613622	Mitochondrial complex I deficiency, nuclear type 19, 618241 (3), Autosomal recessive
<i>FRAS1</i>	607830	Fraser syndrome 1, 219000 (3), Autosomal recessive
<i>FREM1</i>	608944	Manitoba oculotrichoanal syndrome, 248450 (3), Autosomal recessive; Bifid nose with or without anorectal and renal anomalies, 608980 (3), Autosomal recessive; Trigonocephaly 2, 614485 (3), Autosomal dominant
<i>FREM2</i>	608945	Fraser syndrome 2, 617666 (3), Autosomal recessive; Cryptophthalmos, unilateral or bilateral, isolated, 123570 (3), Autosomal recessive
<i>FRMD7</i>	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
<i>FRMPD4</i>	300838	Intellectual developmental disorder, X-linked 104, 300983 (3), X-linked
<i>FRRS1L</i>	604574	Developmental and epileptic encephalopathy 37, 616981 (3), Autosomal recessive
<i>FSHR</i>	136435	Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive
<i>FTCD</i>	606806	Glutamate formiminotransferase deficiency, 229100 (3), Autosomal recessive
<i>FTO</i>	610966	Growth retardation, developmental delay, facial dysmorphism, 612938 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ14}, 612460 (3), Autosomal recessive
<i>FTSJ1</i>	300499	Intellectual developmental disorder, X-linked 9, 309549 (3), X-linked recessive
<i>FUCA1</i>	612280	Fucosidosis, 230000 (3), Autosomal recessive
<i>FYCO1</i>	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
<i>G6PC1</i>	613742	Glycogen storage disease Ia, 232200 (3), Autosomal recessive
<i>G6PC3</i>	611045	Dursun syndrome, 612541 (3), Autosomal recessive; Neutropenia, severe congenital 4, autosomal recessive, 612541 (3), Autosomal recessive
<i>G6PD</i>	305900	Hemolytic anemia, G6PD deficient (favism), 300908 (3), X-linked dominant; {Resistance to malaria due to G6PD deficiency}, 611162 (3)
<i>GAA</i>	606800	Glycogen storage disease II, 232300 (3), Autosomal recessive
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GALE</i>	606953	Galactose epimerase deficiency, 230350 (3), Autosomal recessive

<i>GALK1</i>	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
<i>GALNS</i>	612222	Mucopolysaccharidosis IVA, 253000 (3), Autosomal recessive
<i>GALNT3</i>	601756	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GAMT</i>	601240	Cerebral creatine deficiency syndrome 2, 612736 (3), Autosomal recessive
<i>GAN</i>	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<i>GAS8</i>	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
<i>GATA1</i>	305371	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (3); Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 (3), X-linked recessive; Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 (3), X-linked recessive; Thrombocytopenia with beta-thalassemia, X-linked, 314050 (3), X-linked recessive; Hemolytic anemia due to elevated adenosine deaminase, 301083 (3), X-linked recessive
<i>GATM</i>	602360	Cerebral creatine deficiency syndrome 3, 612718 (3), Autosomal recessive; Fanconi renotubular syndrome 1, 134600 (3), Autosomal dominant
<i>GBA1 (GBA)</i>	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
<i>GBA2</i>	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive
<i>GBE1</i>	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
<i>GCDH</i>	608801	Glutaricaciduria, type I, 231670 (3), Autosomal recessive
<i>GCH1</i>	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
<i>GCK</i>	138079	MODY, type II, 125851 (3), Autosomal dominant; Diabetes mellitus, permanent neonatal 1, 606176 (3), Autosomal recessive; Hyperinsulinemic hypoglycemia, familial, 3, 602485 (3), Autosomal dominant; Diabetes mellitus, noninsulin-dependent, late onset, 125853 (3), Autosomal dominant
<i>GCSH</i>	238330	?Glycine encephalopathy, 605899 (3), Autosomal recessive

<i>GDP1</i>	606598	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate, A, 608340 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2K, 607831 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 4A, 214400 (3), Autosomal recessive
<i>GDF1</i>	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
<i>GDF5</i>	601146	Acromesomelic dysplasia 2A, 200700 (3), Autosomal recessive; Acromesomelic dysplasia 2B, 228900 (3), Autosomal recessive; Multiple synostoses syndrome 2, 610017 (3), Autosomal dominant; Symphalangism, proximal, 1B, 615298 (3), Autosomal dominant; Brachydactyly, type A2, 112600 (3), Autosomal dominant; ?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250 (3), Autosomal recessive; Brachydactyly, type C, 113100 (3), Autosomal dominant; {Osteoarthritis-5}, 612400 (3); Brachydactyly, type A1, C, 615072 (3), Autosomal dominant, Autosomal recessive
<i>GDF6</i>	601147	Microphthalmia with coloboma 6, digenic, 613703 (3), Autosomal dominant; Microphthalmia, isolated 4, 613094 (3); Leber congenital amaurosis 17, 615360 (3), Autosomal recessive; Multiple synostoses syndrome 4, 617898 (3), Autosomal dominant; Klippel-Feil syndrome 1, autosomal dominant, 118100 (3), Autosomal dominant
<i>GDI1</i>	300104	Intellectual developmental disorder, X-linked 41, 300849 (3), X-linked dominant
<i>GFM1</i>	606639	Combined oxidative phosphorylation deficiency 1, 609060 (3), Autosomal recessive
<i>GFPT1</i>	138292	Myasthenia, congenital, 12, with tubular aggregates, 610542 (3), Autosomal recessive
<i>GH1</i>	139250	Kowarski syndrome, 262650 (3), Autosomal recessive; Growth hormone deficiency, isolated, type II, 173100 (3), Autosomal dominant; Growth hormone deficiency, isolated, type IB, 612781 (3); Growth hormone deficiency, isolated, type IA, 262400 (3), Autosomal recessive
<i>GHR</i>	600946	Laron dwarfism, 262500 (3), Autosomal recessive; Increased responsiveness to growth hormone, 604271 (3), Autosomal dominant; Growth hormone insensitivity, partial, 604271 (3), Autosomal dominant; {Hypercholesterolemia, familial, modifier of}, 143890 (3), Autosomal dominant, Autosomal recessive
<i>GHRHR</i>	139191	Growth hormone deficiency, isolated, type IV, 618157 (3), Autosomal recessive
<i>GIPC3</i>	608792	Deafness, autosomal recessive 15, 601869 (3), Autosomal recessive

<i>GJA1</i>	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant
<i>GJB1</i>	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3), X-linked dominant
<i>GJB2</i>	121011	Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant
<i>GJB6</i>	604418	Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive
<i>GJC2</i>	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
<i>GK</i>	300474	Glycerol kinase deficiency, 307030 (3), X-linked recessive
<i>GLA</i>	300644	Fabry disease, cardiac variant, 301500 (3), X-linked; Fabry disease, 301500 (3), X-linked
<i>GLB1</i>	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
<i>GLDC</i>	238300	Glycine encephalopathy, 605899 (3), Autosomal recessive
<i>GLDN</i>	608603	Lethal congenital contracture syndrome 11, 617194 (3), Autosomal recessive
<i>GLE1</i>	603371	Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive
<i>GLIS3</i>	610192	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199 (3), Autosomal recessive
<i>GLRA1</i>	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
<i>GLUL</i>	138290	Glutamine deficiency, congenital, 610015 (3), Autosomal recessive
<i>GLYCTK</i>	610516	D-glycemic aciduria, 220120 (3), Autosomal recessive
<i>GM2A</i>	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive

<i>GMPPA</i>	615495	Alacrima, achalasia, and mental retardation syndrome, 615510 (3), Autosomal recessive
<i>GMPPB</i>	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive
<i>GNAT2</i>	139340	Achromatopsia 4, 613856 (3)
<i>GNB5</i>	604447	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182 (3), Autosomal recessive; Intellectual developmental disorder with cardiac arrhythmia, 617173 (3), Autosomal recessive
<i>GNE</i>	603824	Sialuria, 269921 (3), Autosomal dominant; Nonaka myopathy, 605820 (3), Autosomal recessive
<i>GNMT</i>	606628	Glycine N-methyltransferase deficiency, 606664 (3), Autosomal recessive
<i>GNPAT</i>	602744	Rhizomelic chondrodysplasia punctata, type 2, 222765 (3), Autosomal recessive
<i>GNPTAB</i>	607840	Mucopolysaccharidosis III alpha/beta, 252600 (3), Autosomal recessive; Mucopolysaccharidosis II alpha/beta, 252500 (3), Autosomal recessive
<i>GNPTG</i>	607838	Mucopolysaccharidosis III gamma, 252605 (3), Autosomal recessive
<i>GNRH1</i>	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
<i>GNRHR</i>	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
<i>GNS</i>	607664	Mucopolysaccharidosis type IIID, 252940 (3), Autosomal recessive
<i>GORAB</i>	607983	Geroderma osteodysplasticum, 231070 (3), Autosomal recessive
<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GP1BA</i>	606672	Bernard-Soulier syndrome, type A1 (recessive), 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type A2 (dominant), 153670 (3), Autosomal dominant; von Willebrand disease, platelet-type, 177820 (3), Autosomal dominant; {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 (3), Autosomal recessive
<i>GP1BB</i>	138720	Giant platelet disorder, isolated, 231200 (3), Autosomal recessive; Bernard-Soulier syndrome, type B, 231200 (3), Autosomal recessive
<i>GP9</i>	173515	Bernard-Soulier syndrome, type C, 231200 (3), Autosomal recessive
<i>GPAA1</i>	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
<i>GPC3</i>	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
<i>GPC6</i>	604404	Omodysplasia 1, 258315 (3), Autosomal recessive
<i>GPHN</i>	603930	Molybdenum cofactor deficiency C, 615501 (3), Autosomal recessive
<i>GPI</i>	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470 (3), Autosomal recessive

<i>GPIHBP1</i>	612757	Hyperlipoproteinemia, type 1D, 615947 (3), Autosomal recessive
<i>GPR143</i>	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
<i>GPR179</i>	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
<i>GPSM2</i>	609245	Chudley-McCullough syndrome, 604213 (3), Autosomal recessive
<i>GPT2</i>	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
<i>GRHPR</i>	604296	Hyperoxaluria, primary, type II, 260000 (3), Autosomal recessive
<i>GRIA3</i>	305915	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699 (3), X-linked recessive
<i>GRIP1</i>	604597	Fraser syndrome 3, 617667 (3), Autosomal recessive
<i>GRM1</i>	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
<i>GRM7</i>	604101	Neurodevelopmental disorder with seizures, hypotonia, and brain abnormalities, 618922 (3), Autosomal recessive
<i>GRN</i>	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
<i>GSS</i>	601002	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3), Autosomal recessive; Glutathione synthetase deficiency, 266130 (3), Autosomal recessive
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
<i>GTPBP3</i>	608536	Combined oxidative phosphorylation deficiency 23, 616198 (3), Autosomal recessive
<i>GUCY1A1</i>	139396	Moyamoya 6 with achalasia, 615750 (3), Autosomal recessive
<i>GUCY2C</i>	601330	Diarrhea 6, 614616 (3), Autosomal dominant; Meconium ileus, 614665 (3), Autosomal recessive
<i>GUCY2D</i>	600179	Cone-rod dystrophy 6, 601777 (3), Autosomal dominant, Autosomal recessive; ?Choroidal dystrophy, central areolar 1, 215500 (3), Autosomal dominant; Leber congenital amaurosis 1, 204000 (3), Autosomal recessive; Night blindness, congenital stationary, type 1I, 618555 (3), Autosomal recessive
<i>GUSB</i>	611499	Mucopolysaccharidosis VII, 253220 (3), Autosomal recessive
<i>GYG1</i>	603942	?Glycogen storage disease XV, 613507 (3), Autosomal recessive; Polyglucosan body myopathy 2, 616199 (3), Autosomal recessive
<i>GYS2</i>	138571	Glycogen storage disease 0, liver, 240600 (3), Autosomal recessive
<i>H6PD</i>	138090	Cortisone reductase deficiency 1, 604931 (3), Autosomal recessive
<i>HACE1</i>	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
<i>HADH</i>	601609	Hyperinsulinemic hypoglycemia, familial, 4, 609975 (3), Autosomal recessive; 3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 (3), Autosomal recessive

<i>HADHA</i>	600890	HELLP syndrome, maternal, of pregnancy, 609016 (3), Autosomal recessive; Mitochondrial trifunctional protein deficiency, 609015 (3), Autosomal recessive; LCHAD deficiency, 609016 (3), Autosomal recessive; Fatty liver, acute, of pregnancy, 609016 (3), Autosomal recessive
<i>HADHB</i>	143450	Trifunctional protein deficiency, 609015 (3), Autosomal recessive
<i>HAL</i>	609457	[Histidinemia], 235800 (3), Autosomal dominant, Autosomal recessive
<i>HAMP</i>	606464	Hemochromatosis, type 2B, 613313 (3), Autosomal recessive
<i>HAX1</i>	605998	Neutropenia, severe congenital 3, autosomal recessive, 610738 (3), Autosomal recessive
<i>HBA1</i>	141800	Hemoglobin H disease, nondeletional, 613978 (3); Thalassemias, alpha-, 604131 (3); Heinz body anemias, alpha-, 140700 (3), Autosomal dominant; Methemoglobinemia, alpha type, 617973 (3), Autosomal dominant; Erythrocytosis 7, 617981 (3), Autosomal dominant
<i>HBA2</i>	141850	Heinz body anemia, 140700 (3), Autosomal dominant; Erythrocytosis 7, 617981 (3), Autosomal dominant; Thalassemia, alpha-, 604131 (3); Hemoglobin H disease, deletional and nondeletional, 613978 (3)
<i>HBB</i>	141900	Methemoglobinemia, beta type, 617971 (3), Autosomal dominant; Thalassemia-beta, dominant inclusion-body, 603902 (3), Autosomal dominant; Sickle cell anemia, 603903 (3), Autosomal recessive; Thalassemia, beta, 613985 (3); Delta-beta thalassemia, 141749 (3), Autosomal dominant; {Malaria, resistance to}, 611162 (3); Hereditary persistence of fetal hemoglobin, 141749 (3), Autosomal dominant; Heinz body anemia, 140700 (3), Autosomal dominant; Erythrocytosis 6, 617980 (3), Autosomal dominant
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HCFC1</i>	300019	Methylmalonic aciduria and homocysteinemia, cblX type, 309541 (3), X-linked recessive
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant
<i>HEPACAM</i>	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925 (3), Autosomal recessive; Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926 (3), Autosomal dominant
<i>HERC2</i>	605837	Intellectual developmental disorder, autosomal recessive 38, 615516 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
<i>HES7</i>	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
<i>HESX1</i>	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal dominant, Autosomal recessive; Septo-optic dysplasia, 182230 (3), Autosomal dominant, Autosomal recessive; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal dominant, Autosomal recessive

<i>HEXA</i>	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<i>HGD</i>	607474	Alkaptonuria, 203500 (3), Autosomal recessive
<i>HGSNAT</i>	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930 (3), Autosomal recessive; Retinitis pigmentosa 73, 616544 (3), Autosomal recessive
<i>HIBCH</i>	610690	3-hydroxyisobutryl-CoA hydrolase deficiency, 250620 (3), Autosomal recessive
<i>HINT1</i>	601314	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200 (3), Autosomal recessive
<i>HJV</i>	608374	Hemochromatosis, type 2A, 602390 (3), Autosomal recessive
<i>HK1</i>	142600	Retinitis pigmentosa 79, 617460 (3), Autosomal dominant; Neuropathy, hereditary motor and sensory, Russe type, 605285 (3), Autosomal recessive; Neurodevelopmental disorder with visual defects and brain anomalies, 618547 (3), Autosomal dominant; Hemolytic anemia due to hexokinase deficiency, 235700 (3), Autosomal recessive
<i>HLCS</i>	609018	Holocarboxylase synthetase deficiency, 253270 (3), Autosomal recessive
<i>HMGCL</i>	613898	HMG-CoA lyase deficiency, 246450 (3), Autosomal recessive
<i>HMGCS2</i>	600234	HMG-CoA synthase-2 deficiency, 605911 (3), Autosomal recessive
<i>HNFA1</i>	142410	Hepatic adenoma, somatic, 142330 (3); Diabetes mellitus, insulin-dependent, 20, 612520 (3); {Diabetes mellitus, noninsulin-dependent, 2}, 125853 (3), Autosomal dominant; MODY, type III, 600496 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent}, 222100 (3), Autosomal recessive; Renal cell carcinoma, 144700 (3)
<i>HOGA1</i>	613597	Hyperoxaluria, primary, type III, 613616 (3), Autosomal recessive
<i>HOXA1</i>	142955	Bosley-Salih-Alorainy syndrome, 601536 (3), Autosomal recessive; Athabaskan brainstem dysgenesis syndrome, 601536 (3), Autosomal recessive
<i>HOXA2</i>	604685	Microtia with or without hearing impairment (AD), 612290 (3), Autosomal dominant, Autosomal recessive; ?Microtia, hearing impairment, and cleft palate (AR), 612290 (3), Autosomal dominant, Autosomal recessive
<i>HPD</i>	609695	Hawkinsinuria, 140350 (3), Autosomal dominant; Tyrosinemia, type III, 276710 (3), Autosomal recessive
<i>HPGD</i>	601688	?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
<i>HPRT1</i>	308000	Hyperuricemia, HRPT-related, 300323 (3), X-linked recessive; Lesch-Nyhan syndrome, 300322 (3), X-linked recessive
<i>HPS1</i>	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
<i>HPS3</i>	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive

<i>HPS4</i>	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
<i>HPS5</i>	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
<i>HPS6</i>	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
<i>HPSE2</i>	613469	Urofacial syndrome 1, 236730 (3), Autosomal recessive
<i>HSD11B2</i>	614232	Apparent mineralocorticoid excess, 218030 (3), Autosomal recessive
<i>HSD17B10</i>	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
<i>HSD17B3</i>	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSD3B2</i>	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
<i>HSD3B7</i>	607764	Bile acid synthesis defect, congenital, 1, 607765 (3), Autosomal recessive
<i>HSPD1</i>	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
<i>HSPG2</i>	142461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3), Autosomal recessive; Schwartz-Jampel syndrome, type 1, 255800 (3), Autosomal recessive
<i>HTRA1</i>	602194	{Macular degeneration, age-related, neovascular type}, 610149 (3); {Macular degeneration, age-related, 7}, 610149 (3); CARASIL syndrome, 600142 (3), Autosomal recessive; Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 (3), Autosomal dominant
<i>HTRA2</i>	606441	{Parkinson disease 13}, 610297 (3); 3-methylglutaconic aciduria, type VIII, 617248 (3), Autosomal recessive
<i>HUWE1</i>	300697	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 (3), X-linked
<i>HYAL1</i>	607071	Mucopolysaccharidosis type IX, 601492 (3), Autosomal recessive
<i>HYCC1 (FAM126A)</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>HYDIN</i>	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
<i>HYLS1</i>	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
<i>IARS1</i>	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
<i>IARS2</i>	612801	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007 (3), Autosomal recessive
<i>IBA57</i>	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
<i>ICOS</i>	604558	Immunodeficiency, common variable, 1, 607594 (3), Autosomal recessive
<i>IDH3B</i>	604526	Retinitis pigmentosa 46, 612572 (3), Autosomal recessive

<i>IDS</i>	300823	Mucopolysaccharidosis II, 309900 (3), X-linked recessive
<i>IDUA</i>	252800	Mucopolysaccharidosis Is, 607016 (3), Autosomal recessive; Mucopolysaccharidosis Ih/s, 607015 (3), Autosomal recessive; Mucopolysaccharidosis Ih, 607014 (3), Autosomal recessive
<i>IER3IP1</i>	609382	Microcephaly, epilepsy, and diabetes syndrome, 614231 (3), Autosomal recessive
<i>IFNGR1</i>	107470	{H. pylori infection, susceptibility to}, 600263 (3); Immunodeficiency 27A, mycobacteriosis, AR, 209950 (3), Autosomal recessive; Immunodeficiency 27B, mycobacteriosis, AD, 615978 (3), Autosomal dominant; {Tuberculosis infection, protection against}, 607948 (3); {Tuberculosis, susceptibility to}, 607948 (3); {Hepatitis B virus infection, susceptibility to}, 610424 (3)
<i>IFNGR2</i>	147569	Immunodeficiency 28, mycobacteriosis, 614889 (3), Autosomal recessive
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT140</i>	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT27</i>	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
<i>IFT52</i>	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
<i>IFT80</i>	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
<i>IGBP1</i>	300139	?Corpus callosum, agenesis of, with impaired intellectual development, ocular coloboma and micrognathia, 300472 (3), X-linked recessive
<i>IGF1</i>	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3), Autosomal recessive
<i>IGF1R</i>	147370	Insulin-like growth factor I, resistance to, 270450 (3), Autosomal dominant, Autosomal recessive
<i>IGFBP7</i>	602867	Retinal arterial macroaneurysm with supraaortic pulmonic stenosis, 614224 (3), Autosomal recessive
<i>IGHM</i>	147020	Agammaglobulinemia 1, 601495 (3), Autosomal recessive
<i>IGHMBP2</i>	600502	Neuronopathy, distal hereditary motor, type VI, 604320 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2S, 616155 (3), Autosomal recessive
<i>IHH</i>	600726	Acrocapitofemoral dysplasia, 607778 (3), Autosomal recessive; Brachydactyly, type A1, 112500 (3), Autosomal dominant
<i>IKBKB</i>	603258	Immunodeficiency 15B, 615592 (3), Autosomal recessive; Immunodeficiency 15A, 618204 (3), Autosomal dominant

<i>IKBKG</i>	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
<i>IL10RA</i>	146933	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148 (3), Autosomal recessive
<i>IL11RA</i>	600939	Craniosynostosis and dental anomalies, 614188 (3), Autosomal recessive
<i>IL12B</i>	161561	Immunodeficiency 29, mycobacteriosis, 614890 (3), Autosomal recessive
<i>IL12RB1</i>	601604	Immunodeficiency 30, 614891 (3), Autosomal recessive
<i>IL17RA</i>	605461	Immunodeficiency 51, 613953 (3), Autosomal recessive
<i>IL1RAPL1</i>	300206	Intellectual developmental disorder, X-linked 21, 300143 (3), X-linked recessive
<i>IL1RN</i>	147679	{Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
<i>IL2RG</i>	308380	Combined immunodeficiency, X-linked, moderate, 312863 (3), X-linked recessive; Severe combined immunodeficiency, X-linked, 300400 (3), X-linked recessive
<i>IL36RN</i>	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
<i>IL7R</i>	146661	Immunodeficiency 104, severe combined, 608971 (3), Autosomal recessive
<i>IMPG2</i>	607056	Retinitis pigmentosa 56, 613581 (3), Autosomal recessive; Macular dystrophy, vitelliform, 5, 616152 (3), Autosomal dominant
<i>INPP5E</i>	613037	Joubert syndrome 1, 213300 (3), Autosomal recessive; Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive
<i>INPP5K</i>	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
<i>INPPL1</i>	600829	Opsismodysplasia, 258480 (3), Autosomal recessive
<i>INSR</i>	147670	Rabson-Mendenhall syndrome, 262190 (3), Autosomal recessive; Leprechaunism, 246200 (3), Autosomal recessive; Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (3); Hyperinsulinemic hypoglycemia, familial, 5, 609968 (3), Autosomal dominant
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IQCB1</i>	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
<i>IQSEC2</i>	300522	Intellectual developmental disorder, X-linked 1, 309530 (3), X-linked dominant
<i>ISCA2</i>	615317	Multiple mitochondrial dysfunctions syndrome 4, 616370 (3), Autosomal recessive
<i>ITCH</i>	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive

<i>ITGA2B</i>	607759	Thrombocytopenia, neonatal alloimmune, BAK antigen related (3); Glanzmann thrombasthenia 1, 273800 (3), Autosomal recessive; Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 (3), Autosomal dominant
<i>ITGA6</i>	147556	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817 (3), Autosomal recessive
<i>ITGA8</i>	604063	Renal hypodysplasia/aplasia 1, 191830 (3), Autosomal recessive
<i>ITGB2</i>	600065	Leukocyte adhesion deficiency, 116920 (3), Autosomal recessive
<i>ITGB3</i>	173470	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 (3); Thrombocytopenia, neonatal alloimmune (3); Purpura, posttransfusion (3); {Myocardial infarction, susceptibility to}, 608446 (3); Glanzmann thrombasthenia 2, 619267 (3)
<i>ITGB4</i>	147557	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive
<i>ITK</i>	186973	Lymphoproliferative syndrome 1, 613011 (3), Autosomal recessive
<i>ITPR1</i>	147265	Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant
<i>IVD</i>	607036	Isovaleric acidemia, 243500 (3), Autosomal recessive
<i>IYD</i>	612025	Thyroid dysmorphogenesis 4, 274800 (3), Autosomal recessive
<i>JAGN1</i>	616012	Neutropenia, severe congenital, 6, autosomal recessive, 616022 (3), Autosomal recessive
<i>JAK3</i>	600173	SCID, autosomal recessive, T-negative/B-positive type, 600802 (3), Autosomal recessive
<i>JAM3</i>	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
<i>JUP</i>	173325	Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant
<i>KARS1</i>	601421	Deafness, autosomal recessive 89, 613916 (3), Autosomal recessive; Leukoencephalopathy, progressive, infantile-onset, with or without deafness, 619147 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, recessive intermediate, B, 613641 (3), Autosomal recessive; Deafness, congenital, and adult-onset progressive leukoencephalopathy, 619196 (3), Autosomal recessive
<i>KATNB1</i>	602703	Lissencephaly 6, with microcephaly, 616212 (3), Autosomal recessive
<i>KCNE1</i>	176261	Jervell and Lange-Nielsen syndrome 2, 612347 (3), Autosomal recessive; Long QT syndrome 5, 613695 (3), Autosomal dominant
<i>KCNJ1</i>	600359	Bartter syndrome, type 2, 241200 (3), Autosomal recessive
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive

<i>KCNJ11</i>	600937	Diabetes, permanent neonatal 2, with or without neurologic features, 618856 (3), Autosomal dominant; {Diabetes mellitus, type 2, susceptibility to}, 125853 (3), Autosomal dominant; Maturity-onset diabetes of the young, type 13, 616329 (3), Autosomal dominant; Diabetes mellitus, transient neonatal 3, 610582 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 2, 601820 (3), Autosomal dominant, Autosomal recessive
<i>KCNJ13</i>	603208	Snowflake vitreoretinal degeneration, 193230 (3), Autosomal dominant; Leber congenital amaurosis 16, 614186 (3), Autosomal recessive
<i>KCNQ1</i>	607542	Short QT syndrome 2, 609621 (3), Autosomal dominant; Atrial fibrillation, familial, 3, 607554 (3), Autosomal dominant; Long QT syndrome 1, 192500 (3), Autosomal dominant; {Long QT syndrome 1, acquired, susceptibility to}, 192500 (3), Autosomal dominant; Jervell and Lange-Nielsen syndrome, 220400 (3), Autosomal recessive
<i>KCNV2</i>	607604	Retinal cone dystrophy 3B, 610356 (3), Autosomal recessive
<i>KCTD7</i>	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<i>KDM5C</i>	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
<i>KDM6A</i>	300128	Kabuki syndrome 2, 300867 (3), X-linked dominant
<i>KHDC3L</i>	611687	Hydatidiform mole, recurrent, 2, 614293 (3), Autosomal recessive
<i>KIAA0586</i>	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
<i>KIF14</i>	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
<i>KIF1A</i>	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant, Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal dominant, Autosomal recessive
<i>KIF1C</i>	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
<i>KIF4A</i>	300521	?Intellectual developmental disorder, X-linked 100, 300923 (3), X-linked recessive
<i>KIF7</i>	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydroletharus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
<i>KIFBP</i>	609367	Goldberg-Shprintzen megacolon syndrome, 609460 (3), Autosomal recessive
<i>KISS1R</i>	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
<i>KLHL40</i>	615340	Nemaline myopathy 8, autosomal recessive, 615348 (3), Autosomal recessive

<i>KLHL41</i>	607701	Nemaline myopathy 9, 615731 (3), Autosomal recessive
<i>KLHL7</i>	611119	Retinitis pigmentosa 42, 612943 (3), Autosomal dominant; PERCHING syndrome, 617055 (3), Autosomal recessive
<i>KNL1</i>	609173	Microcephaly 4, primary, autosomal recessive, 604321 (3), Autosomal recessive
<i>KPTN</i>	615620	Intellectual developmental disorder, autosomal recessive 41, 615637 (3), Autosomal recessive
<i>KRT10</i>	148080	Epidermolytic hyperkeratosis, 113800 (3), Autosomal dominant, Autosomal recessive; Ichthyosis with confetti, 609165 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant
<i>KRT14</i>	148066	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex 1C, localized, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex 1A, generalized severe, 131760 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 (3), Autosomal dominant
<i>KRT18</i>	148070	Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive; {Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive
<i>KRT5</i>	148040	Epidermolysis bullosa simplex 2A, generalized severe, 619555 (3), Autosomal dominant; Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 (3), Autosomal dominant; Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 (3), Autosomal recessive; Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 (3), Autosomal dominant; Epidermolysis bullosa simplex 2C, localized, 619594 (3), Autosomal dominant; Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352 (3), Autosomal dominant
<i>KRT8</i>	148060	{Cirrhosis, noncryptogenic, susceptibility to}, 215600 (3), Autosomal recessive; Cirrhosis, cryptogenic, 215600 (3), Autosomal recessive
<i>KRT85</i>	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
<i>KY</i>	605739	Myopathy, myofibrillar, 7, 617114 (3), Autosomal recessive
<i>L1CAM</i>	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; CRASH syndrome, 303350 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive
<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive

<i>LAMA2</i>	156225	Muscular dystrophy, limb-girdle, autosomal recessive 23, 618138 (3), Autosomal recessive; Muscular dystrophy, congenital, merosin deficient or partially deficient, 607855 (3), Autosomal recessive
<i>LAMA3</i>	600805	Epidermolysis bullosa, junctional 2A, intermediate, 619783 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2B, severe, 619784 (3), Autosomal recessive
<i>LAMB1</i>	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<i>LAMB2</i>	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199 (3); Pierson syndrome, 609049 (3), Autosomal recessive
<i>LAMB3</i>	150310	Epidermolysis bullosa, junctional 1B, severe, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional 1A, intermediate, 226650 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant
<i>LAMC2</i>	150292	Epidermolysis bullosa, junctional 3B, severe, 619786 (3), Autosomal recessive; Epidermolysis bullosa, junctional 3A, intermediate, 619785 (3), Autosomal recessive
<i>LAMC3</i>	604349	Cortical malformations, occipital, 614115 (3), Autosomal recessive
<i>LAMP2</i>	309060	Danon disease, 300257 (3), X-linked dominant
<i>LARGE1</i>	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
<i>LARP7</i>	612026	Alazami syndrome, 615071 (3), Autosomal recessive
<i>LARS1</i>	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
<i>LARS2</i>	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
<i>LAS1L</i>	300964	Wilson-Turner syndrome, 309585 (3), X-linked recessive
<i>LAT</i>	602354	Immunodeficiency 52, 617514 (3), Autosomal recessive
<i>LBR</i>	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive
<i>LCA5</i>	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
<i>LCAT</i>	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
<i>LCT</i>	603202	Lactase deficiency, congenital, 223000 (3), Autosomal recessive
<i>LDHA</i>	150000	Glycogen storage disease XI, 612933 (3), Autosomal recessive
<i>LDHB</i>	150100	[Lactate dehydrogenase-B deficiency], 614128 (3)
<i>LDLR</i>	606945	LDL cholesterol level QTL2, 143890 (3), Autosomal dominant, Autosomal recessive; Hypercholesterolemia, familial, 1, 143890 (3), Autosomal dominant, Autosomal recessive
<i>LDLRAP1</i>	605747	Hypercholesterolemia, familial, 4, 603813 (3), Autosomal recessive

<i>LEP</i>	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
<i>LEPR</i>	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive
<i>LGI4</i>	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect, 617468 (3), Autosomal recessive
<i>LHB</i>	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
<i>LHCGR</i>	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
<i>LHX3</i>	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
<i>LIAS</i>	607031	Hyperglycinemia, lactic acidosis, and seizures, 614462 (3), Autosomal recessive
<i>LIFR</i>	151443	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3), Autosomal recessive
<i>LIG4</i>	601837	LIG4 syndrome, 606593 (3), Autosomal recessive; {Multiple myeloma, resistance to}, 254500 (3), Somatic mutation
<i>LINS1</i>	610350	Intellectual developmental disorder, autosomal recessive 27, 614340 (3), Autosomal recessive
<i>LIPA</i>	613497	Wolman disease, 278000 (3), Autosomal recessive; Cholesteryl ester storage disease, 278000 (3), Autosomal recessive
<i>LIPC</i>	151670	{Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant; Hepatic lipase deficiency, 614025 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 12], 612797 (3)
<i>LIPT1</i>	610284	Lipoyltransferase 1 deficiency, 616299 (3), Autosomal recessive
<i>LMAN1</i>	601567	Combined factor V and VIII deficiency, 227300 (3), Autosomal recessive
<i>LMBR1</i>	605522	Triphalangeal thumb, type I, 174500 (3), Autosomal dominant; Syndactyly, type IV, 186200 (3), Autosomal dominant; Laurin-Sandrow syndrome, 135750 (3), Autosomal dominant; Hypoplastic or aplastic tibia with polydactyly, 188740 (3), Autosomal dominant; Polydactyly, preaxial type II, 174500 (3), Autosomal dominant; Acheiropody, 200500 (3), Autosomal recessive; Triphalangeal thumb-polysyndactyly syndrome, 174500 (3), Autosomal dominant
<i>LMBRD1</i>	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive

<i>LMNA</i>	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3); Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
<i>LMOD3</i>	616112	Nemaline myopathy 10, 616165 (3), Autosomal recessive
<i>LONP1</i>	605490	CODAS syndrome, 600373 (3), Autosomal recessive
<i>LOXHD1</i>	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
<i>LPIN1</i>	605518	Myoglobinuria, acute recurrent, autosomal recessive, 268200 (3), Autosomal recessive
<i>LPIN2</i>	605519	Majeed syndrome, 609628 (3)
<i>LPL</i>	609708	Lipoprotein lipase deficiency, 238600 (3), Autosomal recessive; [High density lipoprotein cholesterol level QTL 11], 238600 (3), Autosomal recessive; Combined hyperlipidemia, familial, 144250 (3), Autosomal dominant
<i>LRAT</i>	604863	Leber congenital amaurosis 14, 613341 (3), Autosomal recessive; Retinal dystrophy, early-onset severe, 613341 (3), Autosomal recessive; Retinitis pigmentosa, juvenile, 613341 (3), Autosomal recessive
<i>LRBA</i>	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
<i>LRIG2</i>	608869	Urofacial syndrome 2, 615112 (3), Autosomal recessive
<i>LRMDA</i>	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
<i>LRP2</i>	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
<i>LRP4</i>	604270	?Myasthenic syndrome, congenital, 17, 616304 (3), Autosomal recessive; Sclerosteosis 2, 614305 (3), Autosomal dominant, Autosomal recessive; Cenani-Lenz syndactyly syndrome, 212780 (3), Autosomal recessive
<i>LRP5</i>	603506	Osteopetrosis, autosomal dominant 1, 607634 (3), Autosomal dominant; [Bone mineral density variability 1], 601884 (3), Autosomal dominant; {Osteoporosis}, 166710 (3), Autosomal dominant; Hyperostosis, endosteal, 144750 (3), Autosomal dominant; Osteosclerosis, 144750 (3), Autosomal dominant; Polycystic liver disease 4 with or without kidney cysts, 617875 (3), Autosomal dominant; Osteoporosis-pseudoglioma syndrome, 259770 (3), Autosomal recessive; Exudative vitreoretinopathy 4, 601813 (3), Autosomal dominant, Autosomal recessive; van Buchem disease, type 2, 607636 (3)
<i>LRPPRC</i>	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian), 220111 (3), Autosomal recessive
<i>LRRC51</i>	No OMIM gene	No OMIM phenotype

<i>LRSAM1</i>	610933	Charcot-Marie-Tooth disease, axonal, type 2P, 614436 (3), Autosomal dominant, Autosomal recessive
<i>LRTOMT</i>	612414	Deafness, autosomal recessive 63, 611451 (3), Autosomal recessive
<i>LTBP2</i>	602091	Glaucoma 3, primary congenital, D, 613086 (3); Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750 (3), Autosomal recessive; ?Weill-Marchesani syndrome 3, recessive, 614819 (3), Autosomal recessive
<i>LTBP3</i>	602090	Dental anomalies and short stature, 601216 (3), Autosomal recessive; Geleophysic dysplasia 3, 617809 (3), Autosomal dominant
<i>LTBP4</i>	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
<i>LYRM7</i>	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>LZTR1</i>	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
<i>MAK</i>	154235	Retinitis pigmentosa 62, 614181 (3), Autosomal recessive
<i>MALT1</i>	604860	Immunodeficiency 12, 615468 (3), Autosomal recessive
<i>MAN1B1</i>	604346	Rafiq syndrome, 614202 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MANBA</i>	609489	Mannosidosis, beta, 248510 (3), Autosomal recessive
<i>MAOA</i>	309850	{Antisocial behavior}, 300615 (3), X-linked recessive; Brunner syndrome, 300615 (3), X-linked recessive
<i>MAPKBP1</i>	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
<i>MAPT</i>	157140	Supranuclear palsy, progressive, 601104 (3), Autosomal dominant; Supranuclear palsy, progressive atypical, 260540 (3), Autosomal recessive; Dementia, frontotemporal, with or without parkinsonism, 600274 (3), Autosomal dominant; {Parkinson disease, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant
<i>MARS1</i>	156560	Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
<i>MARS2</i>	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
<i>MASP1</i>	600521	3MC syndrome 1, 257920 (3), Autosomal recessive
<i>MAT1A</i>	610550	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850 (3), Autosomal dominant, Autosomal recessive; Methionine adenosyltransferase deficiency, autosomal recessive, 250850 (3), Autosomal dominant, Autosomal recessive

<i>MBOAT7</i>	606048	Intellectual developmental disorder, autosomal recessive 57, 617188 (3), Autosomal recessive
<i>MBTPS2</i>	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
<i>MC2R</i>	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
<i>MC4R</i>	155541	Obesity (BMIQ20), 618406 (3), Autosomal dominant, Autosomal recessive; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal dominant, Autosomal recessive
<i>MCCC1</i>	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3), Autosomal recessive
<i>MCCC2</i>	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3), Autosomal recessive
<i>MCEE</i>	608419	Methylmalonyl-CoA epimerase deficiency, 251120 (3), Autosomal recessive
<i>MCFD2</i>	607788	Factor V and factor VIII, combined deficiency of, 613625 (3)
<i>MCM4</i>	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
<i>MCOLN1</i>	605248	Mucopolipidosis IV, 252650 (3), Autosomal recessive
<i>MCPH1</i>	607117	Microcephaly 1, primary, autosomal recessive, 251200 (3), Autosomal recessive
<i>MECP2</i>	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
<i>MED12</i>	300188	Lujan-Fryns syndrome, 309520 (3), X-linked recessive; Ohdo syndrome, X-linked, 300895 (3), X-linked recessive; Hardikar syndrome, 301068 (3), X-linked dominant; Opitz-Kaveggia syndrome, 305450 (3), X-linked recessive
<i>MED17</i>	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668 (3), Autosomal recessive
<i>MED23</i>	605042	Intellectual developmental disorder, autosomal recessive 18, with or without epilepsy, 614249 (3), Autosomal recessive
<i>MED25</i>	610197	Basel-Vanagait-Smirin-Yosef syndrome, 616449 (3), Autosomal recessive
<i>MEFV</i>	608107	Neutrophilic dermatosis, acute febrile, 608068 (3), Autosomal dominant; Familial Mediterranean fever, AR, 249100 (3), Autosomal recessive; Familial Mediterranean fever, AD, 134610 (3), Autosomal dominant

<i>MEGF10</i>	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant, 614399 (3), Autosomal recessive; Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399 (3), Autosomal recessive
<i>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<i>MEOX1</i>	600147	Klippel-Feil syndrome 2, 214300 (3), Autosomal recessive
<i>MERTK</i>	604705	Retinitis pigmentosa 38, 613862 (3), Autosomal recessive
<i>MESP2</i>	605195	Spondylocostal dysostosis 2, autosomal recessive, 608681 (3), Autosomal recessive
<i>METTL23</i>	615262	Intellectual developmental disorder, autosomal recessive 44, 615942 (3), Autosomal recessive
<i>MFN2</i>	608507	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
<i>MFRP</i>	606227	Microphthalmia, isolated 5, 611040 (3), Autosomal recessive; Nanophthalmos 2, 609549 (3)
<i>MFSD2A</i>	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities, 616486 (3), Autosomal recessive
<i>MFSD8</i>	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<i>MGAT2</i>	602616	Congenital disorder of glycosylation, type IIa, 212066 (3), Autosomal recessive
<i>MGME1</i>	615076	Mitochondrial DNA depletion syndrome 11, 615084 (3), Autosomal recessive
<i>MGP</i>	154870	Keutel syndrome, 245150 (3), Autosomal recessive
<i>MICU1</i>	605084	Myopathy with extrapyramidal signs, 615673 (3), Autosomal recessive
<i>MID1</i>	300552	Opitz GBBB syndrome, 300000 (3), X-linked recessive
<i>MKKS</i>	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MLH1</i>	120436	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3); Muir-Torre syndrome, 158320 (3), Autosomal dominant; Mismatch repair cancer syndrome 1, 276300 (3), Autosomal recessive
<i>MLPH</i>	606526	GrisCELLI syndrome, type 3, 609227 (3), Autosomal recessive
<i>MLYCD</i>	606761	Malonyl-CoA decarboxylase deficiency, 248360 (3), Autosomal recessive
<i>MMAA</i>	607481	Methylmalonic aciduria, vitamin B12-responsive, cblA type, 251100 (3), Autosomal recessive

<i>MMAB</i>	607568	Methylmalonic aciduria, vitamin B12-responsive, cblB type, 251110 (3), Autosomal recessive
<i>MMACHC</i>	609831	Methylmalonic aciduria and homocystinuria, cblC type, 277400 (3), Autosomal recessive
<i>MMADHC</i>	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
<i>MME</i>	120520	?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal dominant, Autosomal recessive
<i>MMP2</i>	120360	Multicentric osteolysis, nodulosis, and arthropathy, 259600 (3), Autosomal recessive
<i>MMP21</i>	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
<i>MMUT</i>	609058	Methylmalonic aciduria, mut(0) type, 251000 (3), Autosomal recessive
<i>MOCS1</i>	603707	Molybdenum cofactor deficiency A, 252150 (3), Autosomal recessive
<i>MOCS2</i>	603708	Molybdenum cofactor deficiency B, 252160 (3), Autosomal recessive
<i>MOGS</i>	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MPDZ</i>	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219 (3), Autosomal recessive
<i>MPI</i>	154550	Congenital disorder of glycosylation, type Ib, 602579 (3), Autosomal recessive
<i>MPL</i>	159530	Myelofibrosis with myeloid metaplasia, somatic, 254450 (3); Thrombocythemia 2, 601977 (3), Autosomal dominant, Somatic mutation; Thrombocytopenia, congenital amegakaryocytic, 604498 (3), Autosomal recessive
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MPV17</i>	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
<i>MPZ</i>	159440	Charcot-Marie-Tooth disease, type 2I, 607677 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, type 1B, 118200 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, dominant intermediate D, 607791 (3), Autosomal dominant; Hypomyelinating neuropathy, congenital, 2, 618184 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2J, 607736 (3), Autosomal dominant
<i>MRAP</i>	609196	Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive
<i>MRE11</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive

<i>MRPS16</i>	609204	Combined oxidative phosphorylation deficiency 2, 610498 (3), Autosomal recessive
<i>MRPS22</i>	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
<i>MSH2</i>	609309	Muir-Torre syndrome, 158320 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3), Autosomal dominant; Mismatch repair cancer syndrome 2, 619096 (3), Autosomal recessive
<i>MSH6</i>	600678	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 (3), Autosomal dominant; Mismatch repair cancer syndrome 3, 619097 (3), Autosomal recessive; {Endometrial cancer, familial}, 608089 (3), Autosomal dominant, Somatic mutation
<i>MTFMT</i>	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
<i>MTHFD1</i>	172460	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia, 617780 (3), Autosomal recessive
<i>MTHFR</i>	607093	{Vascular disease, susceptibility to} (3); Homocystinuria due to MTHFR deficiency, 236250 (3), Autosomal recessive; {Thromboembolism, susceptibility to}, 188050 (3), Autosomal dominant; {Schizophrenia, susceptibility to}, 181500 (3), Autosomal dominant; {Neural tube defects, susceptibility to}, 601634 (3), Autosomal recessive
<i>MTM1</i>	300415	Myopathy, centronuclear, X-linked, 310400 (3), X-linked recessive
<i>MTMR2</i>	603557	Charcot-Marie-Tooth disease, type 4B1, 601382 (3), Autosomal recessive
<i>MTO1</i>	614667	Combined oxidative phosphorylation deficiency 10, 614702 (3), Autosomal recessive
<i>MTR</i>	156570	{Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive; Homocystinuria-megaloblastic anemia, cblG complementation type, 250940 (3), Autosomal recessive
<i>MTRFR</i>	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<i>MTRR</i>	602568	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3), Autosomal recessive; {Neural tube defects, folate-sensitive, susceptibility to}, 601634 (3), Autosomal recessive
<i>MTTP</i>	157147	{Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant; Abetalipoproteinemia, 200100 (3), Autosomal recessive
<i>MUSK</i>	601296	Fetal akinesia deformation sequence 1, 208150 (3), Autosomal recessive; Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325 (3), Autosomal recessive
<i>MUTYH</i>	604933	Adenomas, multiple colorectal, 608456 (3), Autosomal recessive; Gastric cancer, somatic, 613659 (3)

<i>MVK</i>	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<i>MYBPC3</i>	600958	Cardiomyopathy, hypertrophic, 4, 115197 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1MM, 615396 (3), Autosomal dominant; Left ventricular noncompaction 10, 615396 (3), Autosomal dominant
<i>MYD88</i>	602170	Macroglobulinemia, Waldenstrom, somatic, 153600 (3); Immunodeficiency 68, 612260 (3), Autosomal recessive
<i>MYH2</i>	160740	Proximal myopathy and ophthalmoplegia, 605637 (3), Autosomal dominant, Autosomal recessive
<i>MYH3</i>	160720	Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A, 178110 (3), Autosomal dominant; Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1B, 618469 (3), Autosomal recessive; Arthrogryposis, distal, type 2B3 (Sheldon-Hall), 618436 (3), Autosomal dominant; Arthrogryposis, distal, type 2A (Freeman-Sheldon), 193700 (3), Autosomal dominant
<i>MYH7</i>	160760	Laing distal myopathy, 160500 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 1, 192600 (3), Digenic dominant, Autosomal dominant; Left ventricular noncompaction 5, 613426 (3), Autosomal dominant; Cardiomyopathy, dilated, 1S, 613426 (3), Autosomal dominant; Scapulooperoneal syndrome, myopathic type, 181430 (3), Autosomal dominant; Myopathy, myosin storage, autosomal dominant, 608358 (3), Autosomal dominant; Myopathy, myosin storage, autosomal recessive, 255160 (3), Autosomal recessive
<i>MYMK</i>	615345	Carey-Fineman-Ziter syndrome, 254940 (3), Autosomal recessive
<i>MYO15A</i>	602666	Deafness, autosomal recessive 3, 600316 (3), Autosomal recessive
<i>MYO5A</i>	160777	Griselli syndrome, type 1, 214450 (3), Autosomal recessive
<i>MYO5B</i>	606540	Diarrhea 2, with microvillus atrophy, with or without cholestasis, 251850 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 10, 619868 (3), Autosomal recessive
<i>MYO6</i>	600970	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy, 606346 (3), Autosomal dominant; Deafness, autosomal dominant 22, 606346 (3), Autosomal dominant; Deafness, autosomal recessive 37, 607821 (3), Autosomal recessive
<i>MYO7A</i>	276903	Deafness, autosomal recessive 2, 600060 (3), Autosomal recessive; Usher syndrome, type 1B, 276900 (3), Autosomal recessive; Deafness, autosomal dominant 11, 601317 (3), Autosomal dominant
<i>NAA10</i>	300013	Microphthalmia, syndromic 1, 309800 (3), X-linked; Ogden syndrome, 300855 (3), X-linked recessive, X-linked dominant
<i>NAGA</i>	104170	Schindler disease, type I, 609241 (3), Autosomal recessive; Kanzaki disease, 609242 (3), Autosomal recessive; Schindler disease, type III, 609241 (3), Autosomal recessive

<i>NAGLU</i>	609701	?Charcot-Marie-Tooth disease, axonal, type 2V, 616491 (3), Autosomal dominant; Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920 (3), Autosomal recessive
<i>NAGS</i>	608300	N-acetylglutamate synthase deficiency, 237310 (3), Autosomal recessive
<i>NALCN</i>	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266 (3), Autosomal dominant; Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419 (3), Autosomal recessive
<i>NANS</i>	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
<i>NARS2</i>	612803	Combined oxidative phosphorylation deficiency 24, 616239 (3), Autosomal recessive; ?Deafness, autosomal recessive 94, 618434 (3), Autosomal recessive
<i>NAXE</i>	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
<i>NBAS</i>	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 (3), Autosomal recessive; Infantile liver failure syndrome 2, 616483 (3), Autosomal recessive
<i>NBEAL2</i>	614169	Gray platelet syndrome, 139090 (3), Autosomal recessive
<i>NBN</i>	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
<i>NCF1</i>	608512	Chronic granulomatous disease 1, autosomal recessive, 233700 (3), Autosomal recessive
<i>NCF2</i>	608515	Chronic granulomatous disease 2, autosomal recessive, 233710 (3), Autosomal recessive
<i>NDE1</i>	609449	Microhydranencephaly, 605013 (3), Autosomal recessive; Lissencephaly 4 (with microcephaly), 614019 (3), Autosomal recessive
<i>NDP</i>	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
<i>NDRG1</i>	605262	Charcot-Marie-Tooth disease, type 4D, 601455 (3), Autosomal recessive
<i>NDST1</i>	600853	Intellectual developmental disorder, autosomal recessive 46, 616116 (3), Autosomal recessive
<i>NDUFA1</i>	300078	Mitochondrial complex I deficiency, nuclear type 12, 301020 (3), X-linked recessive
<i>NDUFA10</i>	603835	Mitochondrial complex I deficiency, nuclear type 22, 618243 (3), Autosomal recessive
<i>NDUFA11</i>	612638	Mitochondrial complex I deficiency, nuclear type 14, 618236 (3), Autosomal recessive
<i>NDUFAF2</i>	609653	Mitochondrial complex I deficiency, nuclear type 10, 618233 (3), Autosomal recessive
<i>NDUFAF4</i>	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
<i>NDUFAF5</i>	612360	Mitochondrial complex I deficiency, nuclear type 16, 618238 (3), Autosomal recessive

<i>NDUFAF6</i>	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive
<i>NDUFS1</i>	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
<i>NDUFS2</i>	602985	Mitochondrial complex I deficiency, nuclear type 6, 618228 (3), Autosomal recessive
<i>NDUFS3</i>	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
<i>NDUFS4</i>	602694	Mitochondrial complex I deficiency, nuclear type 1, 252010 (3), Autosomal recessive
<i>NDUFS6</i>	603848	Mitochondrial complex I deficiency, nuclear type 9, 618232 (3), Autosomal recessive
<i>NDUFS7</i>	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
<i>NDUFS8</i>	602141	Mitochondrial complex I deficiency, nuclear type 2, 618222 (3), Autosomal recessive
<i>NDUFV1</i>	161015	Mitochondrial complex I deficiency, nuclear type 4, 618225 (3), Autosomal recessive
<i>NDUFV2</i>	600532	Mitochondrial complex I deficiency, nuclear type 7, 618229 (3), Autosomal recessive
<i>NEB</i>	161650	Nemaline myopathy 2, autosomal recessive, 256030 (3), Autosomal recessive; Arthrogyriposis multiplex congenita 6, 619334 (3), Autosomal recessive
<i>NECTIN1</i>	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
<i>NEFL</i>	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
<i>NEK1</i>	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Digenic recessive, Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
<i>NEK8</i>	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; ?Nephronophthisis 9, 613824 (3)
<i>NEU1</i>	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<i>NEUROG3</i>	604882	Diarrhea 4, malabsorptive, congenital, 610370 (3), Autosomal recessive
<i>NEXMIF</i>	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
<i>NFU1</i>	608100	Multiple mitochondrial dysfunctions syndrome 1, 605711 (3), Autosomal recessive
<i>NGF</i>	162030	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3), Autosomal recessive

<i>NGLY1</i>	610661	Congenital disorder of deglycosylation 1, 615273 (3), Autosomal recessive
<i>NHEJ1</i>	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (3)
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NHS</i>	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
<i>NIN</i>	608684	?Seckel syndrome 7, 614851 (3), Autosomal recessive
<i>NIPAL4</i>	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
<i>NKX2-5</i>	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
<i>NKX3-2</i>	602183	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330 (3), Autosomal recessive
<i>NKX6-2</i>	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
<i>NLGN4X</i>	300427	{Asperger syndrome susceptibility, X-linked 2}, 300497 (3), X-linked; Intellectual developmental disorder, X-linked, 300495 (3), Multifactorial, Isolated cases, X-linked; {Autism susceptibility, X-linked 2}, 300495 (3), Multifactorial, Isolated cases, X-linked
<i>NLRP7</i>	609661	Hydatidiform mole, recurrent, 1, 231090 (3), Autosomal recessive
<i>NMNAT1</i>	608700	Spondyloepiphyseal dysplasia, sensorineural hearing loss, intellectual developmental disorder, and Leber congenital amaurosis, 619260 (3), Autosomal recessive; Leber congenital amaurosis 9, 608553 (3), Autosomal recessive
<i>NNT</i>	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NPHP1</i>	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
<i>NPHP3</i>	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
<i>NPHP4</i>	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
<i>NPHS1</i>	602716	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive

<i>NPHS2</i>	604766	Nephrotic syndrome, type 2, 600995 (3), Autosomal recessive
<i>NPR2</i>	108961	Epiphyseal chondrodysplasia, Miura type, 615923 (3), Autosomal dominant; Short stature with nonspecific skeletal abnormalities, 616255 (3), Autosomal dominant; Acromesomelic dysplasia 1, Maroteaux type, 602875 (3), Autosomal recessive
<i>NR0B1</i>	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
<i>NR2E3</i>	604485	Retinitis pigmentosa 37, 611131 (3), Autosomal dominant, Autosomal recessive; Enhanced S-cone syndrome, 268100 (3), Autosomal recessive
<i>NSDHL</i>	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
<i>NSUN2</i>	610916	Intellectual developmental disorder, autosomal recessive 5, 611091 (3), Autosomal recessive
<i>NT5C2</i>	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
<i>NT5C3A</i>	606224	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3), Autosomal recessive
<i>NTHL1</i>	602656	Familial adenomatous polyposis 3, 616415 (3), Autosomal recessive
<i>NTRK1</i>	191315	Insensitivity to pain, congenital, with anhidrosis, 256800 (3), Autosomal recessive
<i>NUBPL</i>	613621	Mitochondrial complex I deficiency, nuclear type 21, 618242 (3), Autosomal recessive
<i>NUP107</i>	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
<i>NUP62</i>	605815	Striatonigral degeneration, infantile, 271930 (3), Autosomal recessive
<i>NUP93</i>	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
<i>NYX</i>	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
<i>OAT</i>	613349	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870 (3), Autosomal recessive
<i>OBSL1</i>	610991	3-M syndrome 2, 612921 (3), Autosomal recessive
<i>OCA2</i>	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive
<i>OCLN</i>	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<i>OCRL</i>	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
<i>ODAD1</i>	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
<i>ODAD2</i>	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
<i>ODAD3</i>	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive

<i>OFD1</i>	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
<i>OPA1</i>	605290	Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive
<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>OPHN1</i>	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
<i>OPN1LW</i>	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
<i>OPTN</i>	602432	Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435 (3), Autosomal dominant, Autosomal recessive; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
<i>ORAI1</i>	610277	Immunodeficiency 9, 612782 (3), Autosomal recessive; Myopathy, tubular aggregate, 2, 615883 (3), Autosomal dominant
<i>ORC1</i>	601902	Meier-Gorlin syndrome 1, 224690 (3), Autosomal recessive
<i>ORC4</i>	603056	Meier-Gorlin syndrome 2, 613800 (3), Autosomal recessive
<i>ORC6</i>	607213	Meier-Gorlin syndrome 3, 613803 (3), Autosomal recessive
<i>OSGEP</i>	610107	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive
<i>OSTM1</i>	607649	Osteopetrosis, autosomal recessive 5, 259720 (3), Autosomal recessive
<i>OTC</i>	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
<i>OTOA</i>	607038	Deafness, autosomal recessive 22, 607039 (3), Autosomal recessive
<i>OTOF</i>	603681	Auditory neuropathy, autosomal recessive, 1, 601071 (3), Autosomal recessive; Deafness, autosomal recessive 9, 601071 (3), Autosomal recessive
<i>OTOG</i>	604487	Deafness, autosomal recessive 18B, 614945 (3), Autosomal recessive
<i>OTUD6B</i>	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies, 617452 (3), Autosomal recessive
<i>OXCT1</i>	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050 (3), Autosomal recessive
<i>P3H1</i>	610339	Osteogenesis imperfecta, type VIII, 610915 (3), Autosomal recessive
<i>PAH</i>	612349	[Hyperphenylalaninemia, non-PKU mild], 261600 (3), Autosomal recessive; Phenylketonuria, 261600 (3), Autosomal recessive
<i>PAK3</i>	300142	Intellectual developmental disorder, X-linked 30, 300558 (3), X-linked recessive
<i>PANK2</i>	606157	HARP syndrome, 607236 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 1, 234200 (3), Autosomal recessive

<i>PAPSS2</i>	603005	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847 (3), Autosomal recessive
<i>PAX3</i>	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal dominant, Autosomal recessive; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
<i>PAX8</i>	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
<i>PC</i>	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
<i>PCARE</i>	613425	Retinitis pigmentosa 54, 613428 (3)
<i>PCBD1</i>	126090	Hyperphenylalaninemia, BH4-deficient, D, 264070 (3), Autosomal recessive
<i>PCCA</i>	232000	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCCB</i>	232050	Propionicacidemia, 606054 (3), Autosomal recessive
<i>PCDH12</i>	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
<i>PCDH15</i>	605514	Usher syndrome, type 1D/F digenic, 601067 (3), Digenic recessive, Autosomal recessive; Deafness, autosomal recessive 23, 609533 (3), Autosomal recessive; Usher syndrome, type 1F, 602083 (3), Autosomal recessive
<i>PCDH19</i>	300460	Developmental and epileptic encephalopathy 9, 300088 (3), X-linked
<i>PCNT</i>	605925	Microcephalic osteodysplastic primordial dwarfism, type II, 210720 (3), Autosomal recessive
<i>PCSK1</i>	162150	Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ12}, 612362 (3)
<i>PCYT1A</i>	123695	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940 (3), Autosomal recessive
<i>PDE6A</i>	180071	Retinitis pigmentosa 43, 613810 (3)
<i>PDE6B</i>	180072	Retinitis pigmentosa-40, 613801 (3), Autosomal recessive; Night blindness, congenital stationary, autosomal dominant 2, 163500 (3), Autosomal dominant
<i>PDE6C</i>	600827	Cone dystrophy 4, 613093 (3), Autosomal recessive
<i>PDE6D</i>	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive
<i>PDE6H</i>	601190	Retinal cone dystrophy 3, 610024 (3), Autosomal dominant, Autosomal recessive; Achromatopsia 6, 610024 (3), Autosomal dominant, Autosomal recessive
<i>PDHA1</i>	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
<i>PDHB</i>	179060	Pyruvate dehydrogenase E1-beta deficiency, 614111 (3), Autosomal recessive
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PDP1</i>	605993	Pyruvate dehydrogenase phosphatase deficiency, 608782 (3), Autosomal recessive

<i>PDSS1</i>	607429	Coenzyme Q10 deficiency, primary, 2, 614651 (3), Autosomal recessive
<i>PDSS2</i>	610564	Coenzyme Q10 deficiency, primary, 3, 614652 (3), Autosomal recessive
<i>PDX1</i>	600733	{Diabetes mellitus, type II, susceptibility to}, 125853 (3), Autosomal dominant; Pancreatic agenesis 1, 260370 (3), Autosomal recessive; MODY, type IV, 606392 (3)
<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PET100</i>	614770	Mitochondrial complex IV deficiency, nuclear type 12, 619055 (3), Autosomal recessive
<i>PEX1</i>	602136	Heimler syndrome 1, 234580 (3), Autosomal recessive; Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 (3), Autosomal recessive; Peroxisome biogenesis disorder 1A (Zellweger), 214100 (3), Autosomal recessive
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX11B</i>	603867	Peroxisome biogenesis disorder 14B, 614920 (3), Autosomal recessive
<i>PEX12</i>	601758	Peroxisome biogenesis disorder 3B, 266510 (3), Autosomal recessive; Peroxisome biogenesis disorder 3A (Zellweger), 614859 (3), Autosomal recessive
<i>PEX13</i>	601789	Peroxisome biogenesis disorder 11A (Zellweger), 614883 (3), Autosomal recessive; Peroxisome biogenesis disorder 11B, 614885 (3), Autosomal recessive
<i>PEX14</i>	601791	Peroxisome biogenesis disorder 13A (Zellweger), 614887 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
<i>PEX19</i>	600279	Peroxisome biogenesis disorder 12A (Zellweger), 614886 (3), Autosomal recessive
<i>PEX2</i>	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
<i>PEX26</i>	608666	Peroxisome biogenesis disorder 7B, 614873 (3), Autosomal recessive; Peroxisome biogenesis disorder 7A (Zellweger), 614872 (3), Autosomal recessive
<i>PEX3</i>	603164	Peroxisome biogenesis disorder 10A (Zellweger), 614882 (3), Autosomal recessive; ?Peroxisome biogenesis disorder 10B, 617370 (3), Autosomal recessive
<i>PEX5</i>	600414	Peroxisome biogenesis disorder 2B, 202370 (3), Autosomal recessive; Peroxisome biogenesis disorder 2A (Zellweger), 214110 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 5, 616716 (3), Autosomal recessive

<i>PEX6</i>	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
<i>PEX7</i>	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
<i>PFKM</i>	610681	Glycogen storage disease VII, 232800 (3), Autosomal recessive
<i>PGAP1</i>	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
<i>PGAP2</i>	615187	Hyperphosphatasia with mental retardation syndrome 3, 614207 (3), Autosomal recessive
<i>PGAP3</i>	611801	Hyperphosphatasia with mental retardation syndrome 4, 615716 (3), Autosomal recessive
<i>PGK1</i>	311800	Phosphoglycerate kinase 1 deficiency, 300653 (3), X-linked recessive
<i>PGM1</i>	171900	Congenital disorder of glycosylation, type It, 614921 (3), Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PHF6</i>	300414	Borjeson-Forssman-Lehmann syndrome, 301900 (3), X-linked recessive
<i>PHF8</i>	300560	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263 (3), X-linked recessive
<i>PHGDH</i>	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
<i>PHKA2</i>	300798	Glycogen storage disease, type IXa2, 306000 (3), X-linked recessive; Glycogen storage disease, type IXa1, 306000 (3), X-linked recessive
<i>PHKB</i>	172490	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3), Autosomal recessive
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PIEZO1</i>	611184	Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant
<i>PIEZO2</i>	613629	Arthrogryposis, distal, type 5, 108145 (3), Autosomal dominant; Arthrogryposis, distal, with impaired proprioception and touch, 617146 (3), Autosomal recessive; Arthrogryposis, distal, type 3, 114300 (3), Autosomal dominant; ?Marden-Walker syndrome, 248700 (3), Autosomal dominant
<i>PIGA</i>	311770	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 (3); Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 (3), X-linked recessive; Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 (3)
<i>PIGG</i>	616918	[Blood group, EMM system], 619812 (3), Autosomal recessive; Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917 (3), Autosomal recessive

<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive
<i>PIGN</i>	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 (3), Autosomal recessive
<i>PIGO</i>	614730	Hyperphosphatasia with mental retardation syndrome 2, 614749 (3), Autosomal recessive
<i>PIGT</i>	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
<i>PIGV</i>	610274	Hyperphosphatasia with mental retardation syndrome 1, 239300 (3), Autosomal recessive
<i>PINK1</i>	608309	Parkinson disease 6, early onset, 605909 (3), Autosomal recessive
<i>PIP5K1C</i>	606102	Lethal congenital contractural syndrome 3, 611369 (3), Autosomal recessive
<i>PITX3</i>	602669	Cataract 11, multiple types, 610623 (3), Autosomal dominant, Autosomal recessive; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal dominant, Autosomal recessive
<i>PKHD1</i>	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
<i>PKLR</i>	609712	Adenosine triphosphate, elevated, of erythrocytes, 102900 (3), Autosomal dominant; Pyruvate kinase deficiency, 266200 (3), Autosomal recessive
<i>PLA2G6</i>	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
<i>PLAA</i>	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
<i>PLCB4</i>	600810	Auriculocondylar syndrome 2, 614669 (3), Autosomal dominant, Autosomal recessive
<i>PLCE1</i>	608414	Nephrotic syndrome, type 3, 610725 (3), Autosomal recessive
<i>PLEC</i>	601282	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Onga type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
<i>PLEKHG5</i>	611101	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067 (3), Autosomal recessive; Charcot-Marie-Tooth disease, recessive intermediate C, 615376 (3), Autosomal recessive
<i>PLG</i>	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive

<i>PLK4</i>	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171 (3), Autosomal recessive
<i>PLOD1</i>	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 (3), Autosomal recessive
<i>PLOD2</i>	601865	Bruck syndrome 2, 609220 (3), Autosomal recessive
<i>PLOD3</i>	603066	Lysyl hydroxylase 3 deficiency, 612394 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PLPBP</i>	604436	Epilepsy, early-onset, vitamin B6-dependent, 617290 (3), Autosomal recessive
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMP22</i>	601097	Charcot-Marie-Tooth disease, type 1A, 118220 (3), Autosomal dominant; Roussy-Levy syndrome, 180800 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1E, 118300 (3), Autosomal dominant; ?Neuropathy, inflammatory demyelinating, 139393 (3), ?Autosomal dominant; Neuropathy, recurrent, with pressure palsies, 162500 (3), Autosomal dominant; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive
<i>PMPCA</i>	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
<i>PMS2</i>	600259	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 (3); Mismatch repair cancer syndrome 4, 619101 (3), Autosomal recessive
<i>PNKP</i>	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
<i>PNP</i>	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179 (3), Autosomal recessive
<i>PNPLA1</i>	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
<i>PNPLA2</i>	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
<i>PNPLA6</i>	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
<i>PNPO</i>	603287	Pyridoxamine 5'-phosphate oxidase deficiency, 610090 (3), Autosomal recessive
<i>POC1A</i>	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
<i>POLA1</i>	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive

<i>POLG</i>	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POLH</i>	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
<i>POLR1C</i>	610060	Leukodystrophy, hypomyelinating, 11, 616494 (3), Autosomal recessive; Treacher Collins syndrome 3, 248390 (3), Autosomal recessive
<i>POLR3A</i>	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3B</i>	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
<i>POMC</i>	176830	{Obesity, early-onset, susceptibility to}, 601665 (3), Multifactorial, Autosomal dominant, Autosomal recessive; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive
<i>POMGNT1</i>	606822	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
<i>POMGNT2</i>	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
<i>POMK</i>	615247	?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249 (3), Autosomal recessive
<i>POMP</i>	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
<i>POMT1</i>	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155 (3), Autosomal recessive

<i>POMT2</i>	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive
<i>POP1</i>	602486	Anauxetic dysplasia 2, 617396 (3), Autosomal recessive
<i>POR</i>	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>POU1F1</i>	173110	Pituitary hormone deficiency, combined or isolated, 1, 613038 (3), Autosomal dominant, Autosomal recessive
<i>POU3F4</i>	300039	Deafness, X-linked 2, 304400 (3), X-linked recessive
<i>PPA2</i>	609988	?Sudden cardiac failure, alcohol-induced, 617223 (3), Autosomal recessive; Sudden cardiac failure, infantile, 617222 (3), Autosomal recessive
<i>PPIB</i>	123841	Osteogenesis imperfecta, type IX, 259440 (3), Autosomal recessive
<i>PPT1</i>	600722	Ceroid lipofuscinosis, neuronal, 1, 256730 (3), Autosomal recessive
<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRDM12</i>	616458	Neuropathy, hereditary sensory and autonomic, type VIII, 616488 (3), Autosomal recessive
<i>PRDM5</i>	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
<i>PREPL</i>	609557	Myasthenic syndrome, congenital, 22, 616224 (3), Autosomal recessive
<i>PRF1</i>	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<i>PRG4</i>	604283	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3), Autosomal recessive
<i>PRICKLE1</i>	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
<i>PRKN</i>	602544	Adenocarcinoma of lung, somatic, 211980 (3); Parkinson disease, juvenile, type 2, 600116 (3), Autosomal recessive; Ovarian cancer, somatic, 167000 (3)
<i>PROC</i>	612283	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 (3), Autosomal dominant; Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304 (3), Autosomal recessive
<i>PRODH</i>	606810	{Schizophrenia, susceptibility to, 4}, 600850 (3), Autosomal dominant; Hyperprolinemia, type I, 239500 (3), Autosomal recessive
<i>PROM1</i>	604365	Macular dystrophy, retinal, 2, 608051 (3), Autosomal dominant; Retinitis pigmentosa 41, 612095 (3), Autosomal recessive; Stargardt disease 4, 603786 (3), Autosomal dominant; Cone-rod dystrophy 12, 612657 (3), Autosomal dominant, Autosomal recessive
<i>PROP1</i>	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive

<i>PROS1</i>	176880	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 (3), Autosomal recessive; Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336 (3), Autosomal dominant
<i>PRPH2</i>	179605	Macular dystrophy, patterned, 1, 169150 (3), Autosomal dominant; Choroidal dystrophy, central areolar 2, 613105 (3), Autosomal dominant; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Leber congenital amaurosis 18, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive; Macular dystrophy, vitelliform, 3, 608161 (3), Autosomal dominant; Retinitis pigmentosa 7 and digenic form, 608133 (3), Digenic dominant, Autosomal dominant, Autosomal recessive
<i>PRPS1</i>	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
<i>PRRX1</i>	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal dominant, Autosomal recessive
<i>PRSS56</i>	613858	Microphthalmia, isolated 6, 613517 (3), Autosomal recessive
<i>PRUNE1</i>	617413	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies, 617481 (3), Autosomal recessive
<i>PRX</i>	605725	Charcot-Marie-Tooth disease, type 4F, 614895 (3), Autosomal recessive; Dejerine-Sottas disease, 145900 (3), Autosomal dominant, Autosomal recessive
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
<i>PSAT1</i>	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<i>PSMB8</i>	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
<i>PSPH</i>	172480	Phosphoserine phosphatase deficiency, 614023 (3), Autosomal recessive
<i>PTEN</i>	601728	{Glioma susceptibility 2}, 613028 (3); {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
<i>PTH1R</i>	168468	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3), Autosomal dominant; Eiken syndrome, 600002 (3), Autosomal recessive; Failure of tooth eruption, primary, 125350 (3), Autosomal dominant; Chondrodysplasia, Blomstrand type, 215045 (3), Autosomal recessive

<i>PTS</i>	612719	Hyperphenylalaninemia, BH4-deficient, A, 261640 (3), Autosomal recessive
<i>PUS1</i>	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 (3), Autosomal recessive
<i>PXDN</i>	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
<i>PYCR1</i>	179035	Cutis laxa, autosomal recessive, type IIIB, 614438 (3), Autosomal recessive; Cutis laxa, autosomal recessive, type IIB, 612940 (3), Autosomal recessive
<i>PYCR2</i>	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<i>PYGL</i>	613741	Glycogen storage disease VI, 232700 (3), Autosomal recessive
<i>PYGM</i>	608455	McArdle disease, 232600 (3), Autosomal recessive
<i>PYROXD1</i>	617220	Myopathy, myofibrillar, 8, 617258 (3), Autosomal recessive
<i>QARS1</i>	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760 (3), Autosomal recessive
<i>QDPR</i>	612676	Hyperphenylalaninemia, BH4-deficient, C, 261630 (3), Autosomal recessive
<i>RAB18</i>	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<i>RAB23</i>	606144	Carpenter syndrome, 201000 (3), Autosomal recessive
<i>RAB27A</i>	603868	Grisicelli syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAB33B</i>	605950	Smith-McCort dysplasia 2, 615222 (3), Autosomal recessive
<i>RAB39B</i>	300774	Intellectual developmental disorder, X-linked 72, 300271 (3), X-linked recessive; Waisman syndrome, 311510 (3), X-linked recessive
<i>RAB3GAP1</i>	602536	Martsof syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Martsof syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
<i>RAD50</i>	604040	Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive
<i>RAD51C</i>	602774	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 (3); Fanconi anemia, complementation group O, 613390 (3), Autosomal recessive
<i>RAG1</i>	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)
<i>RAG2</i>	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
<i>RAPSN</i>	601592	Fetal akinesia deformation sequence 2, 618388 (3), Autosomal recessive; Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326 (3), Autosomal recessive

<i>RARB</i>	180220	Microphthalmia, syndromic 12, 615524 (3), Autosomal dominant, Autosomal recessive
<i>RARS1</i>	107820	Leukodystrophy, hypomyelinating, 9, 616140 (3), Autosomal recessive
<i>RARS2</i>	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
<i>RAX</i>	601881	Microphthalmia, isolated 3, 611038 (3), Autosomal recessive
<i>RBBP8</i>	604124	Seckel syndrome 2, 606744 (3), Autosomal recessive; Jawad syndrome, 251255 (3), Autosomal recessive; Pancreatic carcinoma, somatic (3)
<i>RBCK1</i>	610924	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895 (3), Autosomal recessive
<i>RBM10</i>	300080	TARP syndrome, 311900 (3), X-linked recessive
<i>RBM8A</i>	605313	Thrombocytopenia-absent radius syndrome, 274000 (3), Autosomal recessive
<i>RCBTB1</i>	607867	Retinal dystrophy with or without extraocular anomalies, 617175 (3), Autosomal recessive
<i>RD3</i>	180040	Leber congenital amaurosis 12, 610612 (3), Autosomal recessive
<i>RDH12</i>	608830	Leber congenital amaurosis 13, 612712 (3), Autosomal dominant, Autosomal recessive
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
<i>REEP1</i>	609139	?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant
<i>REEP6</i>	609346	Retinitis pigmentosa 77, 617304 (3), Autosomal recessive
<i>RELN</i>	600514	{Epilepsy, familial temporal lobe, 7}, 616436 (3), Autosomal dominant; Lissencephaly 2 (Norman-Roberts type), 257320 (3), Autosomal recessive
<i>REN</i>	179820	Renal tubular dysgenesis, 267430 (3), Autosomal recessive; [Hyperproreninemia] (3); Tubulointerstitial kidney disease, autosomal dominant, 4, 613092 (3), Autosomal dominant
<i>RETREG1</i>	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
<i>RFT1</i>	611908	Congenital disorder of glycosylation, type In, 612015 (3), Autosomal recessive
<i>RFX6</i>	612659	Mitchell-Riley syndrome, 615710 (3), Autosomal recessive
<i>RFXANK</i>	603200	MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive
<i>RFXAP</i>	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
<i>RHO</i>	180380	Night blindness, congenital stationary, autosomal dominant 1, 610445 (3), Autosomal dominant; Retinitis pigmentosa 4, autosomal dominant or recessive, 613731 (3), Autosomal dominant, Autosomal recessive; Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive

<i>RIN2</i>	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
<i>RIPK4</i>	605706	CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive
<i>RLBP1</i>	180090	Bothnia retinal dystrophy, 607475 (3), Autosomal recessive; Newfoundland rod-cone dystrophy, 607476 (3); Retinitis punctata albescens, 136880 (3), Autosomal dominant, Autosomal recessive; Fundus albipunctatus, 136880 (3), Autosomal dominant, Autosomal recessive
<i>RLIM</i>	300379	Tonne-Kalscheuer syndrome, 300978 (3), X-linked
<i>RMND1</i>	614917	Combined oxidative phosphorylation deficiency 11, 614922 (3), Autosomal recessive
<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNASF2</i>	612944	Leukoencephalopathy, cystic, without megalencephaly, 612951 (3), Autosomal recessive
<i>RNU4ATAC</i>	601428	Roifman syndrome, 616651 (3), Autosomal recessive; Lowry-Wood syndrome, 226960 (3), Autosomal recessive; Microcephalic osteodysplastic primordial dwarfism, type I, 210710 (3), Autosomal recessive
<i>ROBO3</i>	608630	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313 (3), Autosomal recessive
<i>ROGDI</i>	614574	Kohlschutter-Tonz syndrome, 226750 (3), Autosomal recessive
<i>ROR2</i>	602337	Brachydactyly, type B1, 113000 (3), Autosomal dominant; Robinow syndrome, autosomal recessive, 268310 (3), Autosomal recessive
<i>RORC</i>	602943	Immunodeficiency 42, 616622 (3), Autosomal recessive
<i>RP1</i>	603937	Retinitis pigmentosa 1, 180100 (3), Autosomal dominant, Autosomal recessive
<i>RP2</i>	300757	Retinitis pigmentosa 2, 312600 (3), X-linked
<i>RPE65</i>	180069	Retinitis pigmentosa 20, 613794 (3), Autosomal recessive; Retinitis pigmentosa 87 with choroidal involvement, 618697 (3), Autosomal dominant; Leber congenital amaurosis 2, 204100 (3), Autosomal recessive
<i>RPGR</i>	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
<i>RPGRIP1</i>	605446	Cone-rod dystrophy 13, 608194 (3), Autosomal recessive; Leber congenital amaurosis 6, 613826 (3), Autosomal recessive

<i>RPGRIP1L</i>	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
<i>RPL10</i>	312173	{Autism, susceptibility to, X-linked 5}, 300847 (3); Intellectual developmental disorder, X-linked syndromic 35, 300998 (3), X-linked recessive
<i>RPS6KA3</i>	300075	Intellectual developmental disorder, X-linked 19, 300844 (3), X-linked dominant; Coffin-Lowry syndrome, 303600 (3), X-linked dominant
<i>RRM2B</i>	604712	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 (3), Autosomal recessive; Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 (3), Autosomal dominant
<i>RS1</i>	300839	Retinoschisis, 312700 (3), X-linked recessive
<i>RSPH1</i>	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
<i>RSPH4A</i>	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
<i>RSPH9</i>	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
<i>RTEL1</i>	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive
<i>RTN4IP1</i>	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive
<i>RTTN</i>	610436	Microcephaly, short stature, and polymicrogyria with seizures, 614833 (3), Autosomal recessive
<i>RXYLT1</i>	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
<i>RYR1</i>	180901	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000 (3), Autosomal dominant, Autosomal recessive; Central core disease, 117000 (3), Autosomal dominant, Autosomal recessive; King-Denborough syndrome, 619542 (3), Autosomal dominant; {Malignant hyperthermia susceptibility 1}, 145600 (3), Autosomal dominant; Minicore myopathy with external ophthalmoplegia, 255320 (3), Autosomal recessive
<i>SACS</i>	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
<i>SAMD9</i>	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
<i>SAMHD1</i>	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive

<i>SAR1B</i>	607690	Chylomicron retention disease, 246700 (3), Autosomal recessive
<i>SARS2</i>	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845 (3), Autosomal recessive
<i>SBDS</i>	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
<i>SBF2</i>	607697	Charcot-Marie-Tooth disease, type 4B2, 604563 (3), Autosomal recessive
<i>SC5D</i>	602286	Lathosterolosis, 607330 (3), Autosomal recessive
<i>SCARB2</i>	602257	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900 (3), Autosomal recessive
<i>SCARF2</i>	613619	Van den Ende-Gupta syndrome, 600920 (3), Autosomal recessive
<i>SCN4A</i>	603967	Paramyotonia congenita, 168300 (3), Autosomal dominant; Hypokalemic periodic paralysis, type 2, 613345 (3), Autosomal dominant; Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3), Autosomal dominant; Myasthenic syndrome, congenital, 16, 614198 (3), Autosomal recessive; Hyperkalemic periodic paralysis, type 2, 170500 (3), Autosomal dominant
<i>SCN5A</i>	600163	Ventricular fibrillation, familial, 1, 603829 (3); Heart block, progressive, type IA, 113900 (3), Autosomal dominant; Cardiomyopathy, dilated, 1E, 601154 (3), Autosomal dominant; Heart block, nonprogressive, 113900 (3), Autosomal dominant; Long QT syndrome 3, 603830 (3), Autosomal dominant; Sick sinus syndrome 1, 608567 (3), Autosomal recessive; Brugada syndrome 1, 601144 (3), Autosomal dominant; Atrial fibrillation, familial, 10, 614022 (3), Autosomal dominant; {Sudden infant death syndrome, susceptibility to}, 272120 (3), Autosomal recessive
<i>SCN9A</i>	603415	Erythralgia, primary, 133020 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive
<i>SCNN1A</i>	600228	Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
<i>SCNN1B</i>	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
<i>SCNN1G</i>	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant
<i>SCO1</i>	603644	Mitochondrial complex IV deficiency, nuclear type 4, 619048 (3), Autosomal recessive

<i>SCO2</i>	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
<i>SCYL1</i>	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
<i>SDCCAG8</i>	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Paragangliomas 5, 614165 (3), Autosomal dominant
<i>SDHAF1</i>	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
<i>SDHD</i>	602690	Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive; Pheochromocytoma, 171300 (3), Autosomal dominant
<i>SEC23A</i>	610511	Cranioleptoculosutural dysplasia, 607812 (3), Autosomal dominant, Autosomal recessive
<i>SEC23B</i>	610512	?Cowden syndrome 7, 616858 (3), Autosomal dominant; Dyserythropoietic anemia, congenital, type II, 224100 (3), Autosomal recessive
<i>SELENON</i>	606210	Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal dominant, Autosomal recessive; Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive
<i>SEMA4A</i>	607292	Retinitis pigmentosa 35, 610282 (3), Autosomal dominant, Autosomal recessive; Cone-rod dystrophy 10, 610283 (3), Autosomal recessive
<i>SEPSECS</i>	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<i>SERAC1</i>	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
<i>SERPINA1</i>	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive
<i>SERPINF1</i>	172860	Osteogenesis imperfecta, type VI, 613982 (3), Autosomal recessive
<i>SERPINH1</i>	600943	{Preterm premature rupture of the membranes, susceptibility to}, 610504 (3); Osteogenesis imperfecta, type X, 613848 (3), Autosomal recessive
<i>SETX</i>	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
<i>SFTPB</i>	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive

<i>SGCA</i>	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3, 608099 (3), Autosomal recessive
<i>SGCB</i>	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4, 604286 (3), Autosomal recessive
<i>SGCD</i>	601411	Cardiomyopathy, dilated, 1L, 606685 (3); Muscular dystrophy, limb-girdle, autosomal recessive 6, 601287 (3), Autosomal recessive
<i>SGCG</i>	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5, 253700 (3), Autosomal recessive
<i>SGO1</i>	609168	Chronic atrial and intestinal dysrhythmia, 616201 (3), Autosomal recessive
<i>SGPL1</i>	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
<i>SGSH</i>	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900 (3), Autosomal recessive
<i>SH2D1A</i>	300490	Lymphoproliferative syndrome, X-linked, 1, 308240 (3), X-linked recessive
<i>SH3PXD2B</i>	613293	Frank-ter Haar syndrome, 249420 (3), Autosomal recessive
<i>SH3TC2</i>	608206	Charcot-Marie-Tooth disease, type 4C, 601596 (3), Autosomal recessive; Mononeuropathy of the median nerve, mild, 613353 (3), Autosomal dominant
<i>SHOX</i>	400020	Short stature, idiopathic familial, 300582 (3); Langer mesomelic dysplasia, 249700 (3), Pseudoautosomal recessive; Leri-Weill dyschondrosteosis, 127300 (3), Pseudoautosomal dominant
<i>SHROOM4</i>	300579	Intellectual developmental disorder, X-linked syndromic, Stocco dos Santos type, 300434 (3), X-linked
<i>SI</i>	609845	Sucrase-isomaltase deficiency, congenital, 222900 (3), Autosomal recessive
<i>SIGMAR1</i>	601978	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SIX6</i>	606326	Optic disc anomalies with retinal and/or macular dystrophy, 212550 (3), Autosomal recessive
<i>SKIC2 (SKIV2L)</i>	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
<i>SKIC3 (TTC37)</i>	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
<i>SLC12A1</i>	600839	Bartter syndrome, type 1, 601678 (3), Autosomal recessive
<i>SLC12A3</i>	600968	Gitelman syndrome, 263800 (3), Autosomal recessive
<i>SLC12A5</i>	606726	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685 (3), Autosomal dominant; Developmental and epileptic encephalopathy 34, 616645 (3), Autosomal recessive
<i>SLC12A6</i>	604878	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3), Autosomal recessive
<i>SLC13A5</i>	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive

<i>SLC16A1</i>	600682	Hyperinsulinemic hypoglycemia, familial, 7, 610021 (3), Autosomal dominant; Erythrocyte lactate transporter defect, 245340 (3), Autosomal dominant; Monocarboxylate transporter 1 deficiency, 616095 (3), Autosomal dominant, Autosomal recessive
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC19A2</i>	603941	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3), Autosomal recessive
<i>SLC19A3</i>	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
<i>SLC1A4</i>	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
<i>SLC22A12</i>	607096	Hypouricemia, renal, 220150 (3), Autosomal recessive
<i>SLC22A5</i>	603377	Carnitine deficiency, systemic primary, 212140 (3), Autosomal recessive
<i>SLC24A5</i>	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
<i>SLC25A1</i>	190315	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182 (3), Autosomal recessive; Myasthenic syndrome, congenital, 23, presynaptic, 618197 (3), Autosomal recessive
<i>SLC25A13</i>	603859	Citrullinemia, type II, neonatal-onset, 605814 (3), Autosomal recessive; Citrullinemia, adult-onset type II, 603471 (3), Autosomal recessive
<i>SLC25A15</i>	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
<i>SLC25A19</i>	606521	Microcephaly, Amish type, 607196 (3), Autosomal recessive; Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710 (3), Autosomal recessive
<i>SLC25A20</i>	613698	Carnitine-acylcarnitine translocase deficiency, 212138 (3), Autosomal recessive
<i>SLC25A22</i>	609302	Developmental and epileptic encephalopathy 3, 609304 (3), Autosomal recessive
<i>SLC25A38</i>	610819	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 (3), Autosomal recessive
<i>SLC25A4</i>	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184 (3), Autosomal dominant
<i>SLC25A46</i>	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive

<i>SLC26A2</i>	606718	Epiphyseal dysplasia, multiple, 4, 226900 (3), Autosomal recessive; De la Chapelle dysplasia, 256050 (3), Autosomal recessive; Diastrophic dysplasia, 222600 (3), Autosomal recessive; Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 (3), Autosomal recessive; Achondrogenesis Ib, 600972 (3), Autosomal recessive; Atelosteogenesis, type II, 256050 (3), Autosomal recessive
<i>SLC26A3</i>	126650	Diarrhea 1, secretory chloride, congenital, 214700 (3), Autosomal recessive
<i>SLC26A4</i>	605646	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791 (3), Autosomal recessive; Pendred syndrome, 274600 (3), Autosomal recessive
<i>SLC27A4</i>	604194	Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive
<i>SLC29A3</i>	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant
<i>SLC2A10</i>	606145	Arterial tortuosity syndrome, 208050 (3), Autosomal recessive
<i>SLC2A2</i>	138160	Fanconi-Bickel syndrome, 227810 (3), Autosomal recessive; {Diabetes mellitus, noninsulin-dependent}, 125853 (3), Autosomal dominant
<i>SLC2A9</i>	606142	{Uric acid concentration, serum, QTL 2}, 612076 (3), Autosomal dominant, Autosomal recessive; Hypouricemia, renal, 2, 612076 (3), Autosomal dominant, Autosomal recessive
<i>SLC30A10</i>	611146	Hypermanganesemia with dystonia 1, 613280 (3), Autosomal recessive
<i>SLC33A1</i>	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive
<i>SLC34A2</i>	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
<i>SLC35A1</i>	605634	Congenital disorder of glycosylation, type II _f , 603585 (3), Autosomal recessive
<i>SLC35A2</i>	314375	Congenital disorder of glycosylation, type II _m , 300896 (3), Somatic mosaicism, X-linked dominant
<i>SLC35A3</i>	605632	Arthrogryposis, impaired intellectual development, and seizures, 615553 (3), Autosomal recessive
<i>SLC35C1</i>	605881	Congenital disorder of glycosylation, type II _c , 266265 (3), Autosomal recessive
<i>SLC35D1</i>	610804	Schneckenbecken dysplasia, 269250 (3), Autosomal recessive
<i>SLC37A4</i>	602671	Glycogen storage disease Ib, 232220 (3), Autosomal recessive; Congenital disorder of glycosylation, type II _w , 619525 (3), Autosomal dominant; Glycogen storage disease Ic, 232240 (3), Autosomal recessive

<i>SLC38A8</i>	615585	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218 (3), Autosomal recessive
<i>SLC39A14</i>	608736	?Hyperostosis cranialis interna, 144755 (3), Autosomal dominant; Hypermanganemia with dystonia 2, 617013 (3), Autosomal recessive
<i>SLC39A4</i>	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
<i>SLC39A8</i>	608732	Congenital disorder of glycosylation, type II _n , 616721 (3), Autosomal recessive
<i>SLC3A1</i>	104614	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive
<i>SLC45A2</i>	606202	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive
<i>SLC46A1</i>	611672	Folate malabsorption, hereditary, 229050 (3), Autosomal recessive
<i>SLC4A1</i>	109270	[Blood group, Swann], 601550 (3); [Blood group, Wright], 112050 (3); Distal renal tubular acidosis 1, 179800 (3), Autosomal dominant; [Blood group, Waldner], 112010 (3); Spherocytosis, type 4, 612653 (3), Autosomal dominant; [Blood group, Froese], 601551 (3); Distal renal tubular acidosis 4 with hemolytic anemia, 611590 (3), Autosomal recessive; {Malaria, resistance to}, 611162 (3); Cryohydrocytosis, 185020 (3), Autosomal dominant; Ovalocytosis, SA type, 166900 (3), Autosomal dominant; [Blood group, Diego], 110500 (3)
<i>SLC4A11</i>	610206	Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive; Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive
<i>SLC4A4</i>	603345	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3), Autosomal recessive
<i>SLC52A2</i>	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
<i>SLC52A3</i>	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
<i>SLC5A5</i>	601843	Thyroid dysmorphogenesis 1, 274400 (3), Autosomal recessive
<i>SLC5A7</i>	608761	Neuronopathy, distal hereditary motor, type VIIA, 158580 (3), Autosomal dominant; Myasthenic syndrome, congenital, 20, presynaptic, 617143 (3), Autosomal recessive
<i>SLC6A19</i>	608893	Iminoglycinuria, digenic, 242600 (3), Digenic recessive, Autosomal recessive; Hartnup disorder, 234500 (3), Autosomal recessive; Hyperglycinuria, 138500 (3), Autosomal dominant
<i>SLC6A3</i>	126455	Parkinsonism-dystonia, infantile, 1, 613135 (3), Autosomal recessive; {Nicotine dependence, protection against}, 188890 (3)
<i>SLC6A5</i>	604159	Hyperekplexia 3, 614618 (3), Autosomal dominant, Autosomal recessive
<i>SLC6A8</i>	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
<i>SLC7A7</i>	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
<i>SLC7A9</i>	604144	Cystinuria, 220100 (3), Autosomal dominant, Autosomal recessive

<i>SLC9A3</i>	182307	Diarrhea 8, secretory sodium, congenital, 616868 (3), Autosomal recessive
<i>SLC9A6</i>	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked
<i>SLX4</i>	613278	Fanconi anemia, complementation group P, 613951 (3), Autosomal recessive
<i>SMARCAL1</i>	606622	Schimke immunoosseous dysplasia, 242900 (3), Autosomal recessive
<i>SMC1A</i>	300040	Cornelia de Lange syndrome 2, 300590 (3), X-linked dominant; Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 (3), X-linked dominant
<i>SMN1</i>	600354	Spinal muscular atrophy-2, 253550 (3), Autosomal recessive; Spinal muscular atrophy-4, 271150 (3), Autosomal recessive; Spinal muscular atrophy-3, 253400 (3), Autosomal recessive; Spinal muscular atrophy-1, 253300 (3), Autosomal recessive
<i>SMN2</i>	601627	{Spinal muscular atrophy, type III, modifier of}, 253400 (3), Autosomal recessive
<i>SMOC1</i>	608488	Microphthalmia with limb anomalies, 206920 (3), Autosomal recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
<i>SMS</i>	300105	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583 (3), X-linked recessive
<i>SNAP29</i>	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
<i>SNIP1</i>	608241	Neurodevelopmental disorder with hypotonia, craniofacial abnormalities, and seizures, 614501 (3), Autosomal recessive
<i>SNORD118</i>	616663	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive
<i>SNX14</i>	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<i>SOD1</i>	147450	Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal dominant, Autosomal recessive
<i>SOST</i>	605740	Sclerosteosis 1, 269500 (3), Autosomal recessive; Craniodiaphyseal dysplasia, autosomal dominant, 122860 (3), Autosomal dominant
<i>SOX3</i>	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
<i>SP110</i>	604457	{Mycobacterium tuberculosis, susceptibility to}, 607948 (3); Hepatic venoocclusive disease with immunodeficiency, 235550 (3), Autosomal recessive
<i>SPAG1</i>	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
<i>SPART</i>	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<i>SPATA5</i>	613940	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577 (3), Autosomal recessive

<i>SPATA7</i>	609868	Leber congenital amaurosis 3, 604232 (3), Autosomal recessive; Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 (3), Autosomal recessive
<i>SPG11</i>	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
<i>SPG7</i>	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive
<i>SPINK5</i>	605010	Netherton syndrome, 256500 (3), Autosomal recessive
<i>SPINT2</i>	605124	Diarrhea 3, secretory sodium, congenital, syndromic, 270420 (3), Autosomal recessive
<i>SPR</i>	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
<i>SPTA1</i>	182860	Spherocytosis, type 3, 270970 (3), Autosomal recessive; Elliptocytosis-2, 130600 (3), Autosomal dominant; Pyropoikilocytosis, 266140 (3), Autosomal recessive
<i>SPTBN2</i>	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
<i>SQSTM1</i>	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
<i>SRD5A2</i>	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
<i>SRD5A3</i>	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
<i>SSR4</i>	300090	Congenital disorder of glycosylation, type Iy, 300934 (3), X-linked recessive
<i>ST3GAL3</i>	606494	Developmental and epileptic encephalopathy 15, 615006 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 12, 611090 (3), Autosomal recessive
<i>ST3GAL5</i>	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
<i>STAC3</i>	615521	Myopathy, congenital, Baily-Bloch, 255995 (3), Autosomal recessive
<i>STAMBP</i>	606247	Microcephaly-capillary malformation syndrome, 614261 (3), Autosomal recessive
<i>STAR</i>	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive

<i>STAT1</i>	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant, 614162 (3), Autosomal dominant; Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892 (3), Autosomal dominant; Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796 (3), Autosomal recessive
<i>STIL</i>	181590	Microcephaly 7, primary, autosomal recessive, 612703 (3), Autosomal recessive
<i>STIM1</i>	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
<i>STRA6</i>	610745	Microphthalmia, syndromic 9, 601186 (3), Autosomal recessive; Microphthalmia, isolated, with coloboma 8, 601186 (3), Autosomal recessive
<i>STRADA</i>	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087 (3), Autosomal recessive
<i>STRC</i>	606440	Deafness, autosomal recessive 16, 603720 (3), Autosomal recessive
<i>STS</i>	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
<i>STUB1</i>	607207	Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
<i>STX11</i>	605014	Hemophagocytic lymphohistiocytosis, familial, 4, 603552 (3), Autosomal recessive
<i>STXBP2</i>	601717	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101 (3)
<i>SUCLA2</i>	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<i>SUCLG1</i>	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400 (3), Autosomal recessive
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>SURF1</i>	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
<i>SYN1</i>	313440	Intellectual developmental disorder, X-linked 50, 300115 (3), X-linked; Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3), X-linked recessive, X-linked dominant
<i>SYNE1</i>	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
<i>SYP</i>	313475	Intellectual developmental disorder, X-linked 96, 300802 (3), X-linked recessive
<i>SZT2</i>	615463	Developmental and epileptic encephalopathy 18, 615476 (3), Autosomal recessive

<i>TACR3</i>	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
<i>TACSTD2</i>	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
<i>TAF2</i>	604912	Intellectual developmental disorder, autosomal recessive 40, 615599 (3), Autosomal recessive
<i>TAFAZZIN</i>	300394	Barth syndrome, 302060 (3), X-linked recessive
<i>TALDO1</i>	602063	Transaldolase deficiency, 606003 (3), Autosomal recessive
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TAP1</i>	170260	Bare lymphocyte syndrome, type I, 604571 (3), Autosomal recessive
<i>TAT</i>	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TBC1D23</i>	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
<i>TBC1D24</i>	613577	Deafness, autosomal recessive 86, 614617 (3), Autosomal recessive; Epilepsy, rolandic, with paroxysmal exercise-induced dystonia and writer's cramp, 608105 (3), Autosomal recessive; Myoclonic epilepsy, infantile, familial, 605021 (3), Autosomal recessive; Deafness, autosomal dominant 65, 616044 (3), Autosomal dominant; Developmental and epileptic encephalopathy 16, 615338 (3), Autosomal recessive; DOORS syndrome, 220500 (3), Autosomal recessive
<i>TBCD</i>	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193 (3), Autosomal recessive
<i>TBCE</i>	604934	Kenny-Caffey syndrome, type 1, 244460 (3), Autosomal recessive; Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3), Autosomal recessive; Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 (3), Autosomal recessive
<i>TBCK</i>	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900 (3), Autosomal recessive
<i>TBX15</i>	604127	Cousin syndrome, 260660 (3), Autosomal recessive
<i>TBX19</i>	604614	Adrenocorticotrophic hormone deficiency, 201400 (3), Autosomal recessive
<i>TBX22</i>	300307	Cleft palate with ankyloglossia, 303400 (3), X-linked; ?Abruzzo-Erickson syndrome, 302905 (3), X-linked
<i>TBX6</i>	602427	Spondylocostal dysostosis 5, 122600 (3), Autosomal dominant, Autosomal recessive
<i>TBXAS1</i>	274180	Ghosal hematodiaphyseal syndrome, 231095 (3), Autosomal recessive
<i>TCAP</i>	604488	Cardiomyopathy, hypertrophic, 25, 607487 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 7, 601954 (3), Autosomal recessive
<i>TCIRG1</i>	604592	Osteopetrosis, autosomal recessive 1, 259700 (3), Autosomal recessive
<i>TCN2</i>	613441	Transcobalamin II deficiency, 275350 (3), Autosomal recessive
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive

<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
<i>TDRD7</i>	611258	Cataract 36, 613887 (3), Autosomal recessive
<i>TECPR2</i>	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
<i>TECTA</i>	602574	Deafness, autosomal dominant 8/12, 601543 (3), Autosomal dominant; Deafness, autosomal recessive 21, 603629 (3), Autosomal recessive
<i>TELO2</i>	611140	You-Hoover-Fong syndrome, 616954 (3), Autosomal recessive
<i>TERT</i>	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal dominant, Autosomal recessive; {Melanoma, cutaneous malignant, 9}, 615134 (3); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation
<i>TF</i>	190000	Atransferrinemia, 209300 (3), Autosomal recessive
<i>TFR2</i>	604720	Hemochromatosis, type 3, 604250 (3), Autosomal recessive
<i>TG</i>	188450	{Autoimmune thyroid disease, susceptibility to, 3}, 608175 (3); Thyroid dyshormonogenesis 3, 274700 (3), Autosomal recessive
<i>TGM1</i>	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
<i>TH</i>	191290	Segawa syndrome, recessive, 605407 (3), Autosomal recessive
<i>THOC2</i>	300395	Intellectual developmental disorder, X-linked 12, 300957 (3), X-linked recessive
<i>THRB</i>	190160	Thyroid hormone resistance, autosomal recessive, 274300 (3), Autosomal recessive; Thyroid hormone resistance, 188570 (3), Autosomal dominant; Thyroid hormone resistance, selective pituitary, 145650 (3), Autosomal dominant
<i>TIMM8A</i>	300356	Mohr-Tranebjaerg syndrome, 304700 (3), X-linked recessive
<i>TJP2</i>	607709	Hypercholanemia, familial 1, 607748 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic 4, 615878 (3), Autosomal recessive
<i>TK2</i>	188250	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560 (3), Autosomal recessive; ?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069 (3), Autosomal recessive
<i>TMC1</i>	606706	Deafness, autosomal dominant 36, 606705 (3), Autosomal dominant; Deafness, autosomal recessive 7, 600974 (3), Autosomal recessive
<i>TMCO1</i>	614123	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980 (3), Autosomal recessive

<i>TMEM107</i>	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
<i>TMEM126A</i>	612988	Optic atrophy 7, 612989 (3), Autosomal recessive
<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM165</i>	614726	Congenital disorder of glycosylation, type IIk, 614727 (3), Autosomal recessive
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM67</i>	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
<i>TMEM70</i>	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052 (3), Autosomal recessive
<i>TMIE</i>	607237	Deafness, autosomal recessive 6, 600971 (3), Autosomal recessive
<i>TMPRSS3</i>	605511	Deafness, autosomal recessive 8/10, 601072 (3), Autosomal recessive
<i>TMTC3</i>	617218	Lissencephaly 8, 617255 (3), Autosomal recessive
<i>TNFRSF11A</i>	603499	Osteopetrosis, autosomal recessive 7, 612301 (3), Autosomal recessive; {Paget disease of bone 2, early-onset}, 602080 (3), Autosomal dominant; Osteolysis, familial expansile, 174810 (3), Autosomal dominant
<i>TNFRSF11B</i>	602643	Paget disease of bone 5, juvenile-onset, 239000 (3), Autosomal recessive
<i>TNFRSF13B</i>	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal dominant, Autosomal recessive; Immunoglobulin A deficiency 2, 609529 (3)
<i>TNFSF11</i>	602642	Osteopetrosis, autosomal recessive 2, 259710 (3), Autosomal recessive
<i>TNNI3</i>	191044	?Cardiomyopathy, dilated, 2A, 611880 (3), Autosomal recessive; Cardiomyopathy, hypertrophic, 7, 613690 (3), Autosomal dominant; Cardiomyopathy, familial restrictive, 1, 115210 (3), Autosomal dominant; Cardiomyopathy, dilated, 1FF, 613286 (3)
<i>TNNT1</i>	191041	Nemaline myopathy 5, Amish type, 605355 (3), Autosomal recessive
<i>TNXB</i>	600985	Ehlers-Danlos syndrome, classic-like, 1, 606408 (3), Autosomal recessive; Vesicoureteral reflux 8, 615963 (3), Autosomal dominant
<i>TOE1</i>	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
<i>TPI1</i>	190450	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512 (3), Autosomal recessive
<i>TPK1</i>	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458 (3), Autosomal recessive

<i>TPM3</i>	191030	CAP myopathy 1, 609284 (3), Autosomal dominant, Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal dominant, Autosomal recessive; Nemaline myopathy 1, autosomal dominant or recessive, 609284 (3), Autosomal dominant, Autosomal recessive
<i>TPO</i>	606765	Thyroid dysmorphogenesis 2A, 274500 (3), Autosomal recessive
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TRAC</i>	186880	Immunodeficiency 7, TCR-alpha/beta deficient, 615387 (3), Autosomal recessive
<i>TRAPPC11</i>	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18, 615356 (3), Autosomal recessive
<i>TRAPPC9</i>	611966	Intellectual developmental disorder, autosomal recessive 13, 613192 (3), Autosomal recessive
<i>TRDN</i>	603283	Cardiac arrhythmia syndrome, with or without skeletal muscle weakness, 615441 (3), Autosomal recessive
<i>TREM2</i>	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193 (3), Autosomal recessive
<i>TREX1</i>	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal dominant, Autosomal recessive; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TRIM37</i>	605073	Mulibrey nanism, 253250 (3), Autosomal recessive
<i>TRIOBP</i>	609761	Deafness, autosomal recessive 28, 609823 (3), Autosomal recessive
<i>TRIP11</i>	604505	Odontochondrodysplasia 1, 184260 (3), Autosomal recessive; Achondrogenesis, type IA, 200600 (3), Autosomal recessive
<i>TRIT1</i>	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
<i>TRMT10A</i>	616013	Microcephaly, short stature, and impaired glucose metabolism 1, 616033 (3), Autosomal recessive
<i>TRMU</i>	610230	{Deafness, mitochondrial, modifier of}, 580000 (3), Mitochondrial; Liver failure, transient infantile, 613070 (3), Autosomal recessive
<i>TRNT1</i>	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
<i>TRPM6</i>	607009	Hypomagnesemia 1, intestinal, 602014 (3), Autosomal recessive
<i>TSEN2</i>	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive

<i>TSEN34</i>	608754	?Pontocerebellar hypoplasia type 2C, 612390 (3), Autosomal recessive
<i>TSEN54</i>	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<i>TSMF</i>	604723	Combined oxidative phosphorylation deficiency 3, 610505 (3), Autosomal recessive
<i>TSHB</i>	188540	Hypothyroidism, congenital, nongoitrous 4, 275100 (3), Autosomal recessive
<i>TSHR</i>	603372	Hyperthyroidism, familial gestational, 603373 (3), Autosomal dominant; Hyperthyroidism, nonautoimmune, 609152 (3), Autosomal dominant; Thyroid adenoma, hyperfunctioning, somatic (3); Hypothyroidism, congenital, nongoitrous, 1, 275200 (3), Autosomal recessive; Thyroid carcinoma with thyrotoxicosis, somatic (3)
<i>TSPAN7</i>	300096	Intellectual developmental disorder, X-linked 58, 300210 (3), X-linked recessive
<i>TSPYL1</i>	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
<i>TTC19</i>	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
<i>TTC21B</i>	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal dominant, Autosomal recessive
<i>TTC7A</i>	609332	Gastrointestinal defects and immunodeficiency syndrome, 243150 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TTI2</i>	614426	Intellectual developmental disorder, autosomal recessive 39, 615541 (3), Autosomal recessive
<i>TTN</i>	188840	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807 (3), Autosomal recessive; Cardiomyopathy, familial hypertrophic, 9, 613765 (3), Autosomal dominant; Tibial muscular dystrophy, tardive, 600334 (3), Autosomal dominant; Salih myopathy, 611705 (3), Autosomal recessive; Cardiomyopathy, dilated, 1G, 604145 (3), Autosomal dominant; Myopathy, myofibrillar, 9, with early respiratory failure, 603689 (3), Autosomal dominant
<i>TTPA</i>	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<i>TUBA8</i>	605742	Macrothrombocytopenia, isolated, 2, autosomal dominant, 619840 (3), Autosomal dominant
<i>TUBGCP4</i>	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>TUBGCP6</i>	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270 (3), Autosomal recessive

<i>TUFM</i>	602389	Combined oxidative phosphorylation deficiency 4, 610678 (3), Autosomal recessive
<i>TULP1</i>	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
<i>TUSC3</i>	601385	Intellectual developmental disorder, autosomal recessive 7, 611093 (3), Autosomal recessive
<i>TWNK</i>	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
<i>TXNL4A</i>	611595	Burn-McKeown syndrome, 608572 (3), Autosomal recessive
<i>TYK2</i>	176941	Immunodeficiency 35, 611521 (3), Autosomal recessive
<i>TYMP</i>	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041 (3), Autosomal recessive
<i>TYR</i>	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (3), Autosomal dominant; [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800 (3), Autosomal dominant; Albinism, oculocutaneous, type IB, 606952 (3), Autosomal recessive; Waardenburg syndrome/albinism, digenic, 103470 (3); Albinism, oculocutaneous, type IA, 203100 (3), Autosomal recessive
<i>TYROBP</i>	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770 (3), Autosomal recessive
<i>TYRP1</i>	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive
<i>UBA1</i>	314370	Spinal muscular atrophy, X-linked 2, infantile, 301830 (3), X-linked recessive; VEXAS syndrome, somatic, 301054 (3)
<i>UBA5</i>	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
<i>UBE2A</i>	312180	Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 (3), X-linked recessive
<i>UBE2T</i>	610538	Fanconi anemia, complementation group T, 616435 (3), Autosomal recessive
<i>UBE3B</i>	608047	Kaufman oculocerebrofacial syndrome, 244450 (3), Autosomal recessive
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>UFM1</i>	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
<i>UGT1A1</i>	191740	Crigler-Najjar syndrome, type I, 218800 (3), Autosomal recessive; [Bilirubin, serum level of, QTL1], 601816 (3); Hyperbilirubinemia, familial transient neonatal, 237900 (3), Autosomal dominant, Autosomal recessive; Crigler-Najjar syndrome, type II, 606785 (3), Autosomal recessive; [Gilbert syndrome], 143500 (3), Autosomal recessive

<i>UMPS</i>	613891	Orotic aciduria, 258900 (3), Autosomal recessive
<i>UNC13D</i>	608897	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3), Autosomal recessive
<i>UNC80</i>	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801 (3), Autosomal recessive
<i>UPB1</i>	606673	Beta-ureidopropionase deficiency, 613161 (3), Autosomal recessive
<i>UPF3B</i>	300298	Intellectual developmental disorder, X-linked syndromic 14, 300676 (3), X-linked recessive
<i>UQCRB</i>	191330	Mitochondrial complex III deficiency, nuclear type 3, 615158 (3), Autosomal recessive
<i>UQCRC2</i>	191329	Mitochondrial complex III deficiency, nuclear type 5, 615160 (3), Autosomal recessive
<i>UQCRCQ</i>	612080	Mitochondrial complex III deficiency, nuclear type 4, 615159 (3), Autosomal recessive
<i>UROS</i>	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
<i>USB1</i>	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
<i>USH1C</i>	605242	Usher syndrome, type 1C, 276904 (3), Autosomal recessive; Deafness, autosomal recessive 18A, 602092 (3), Autosomal recessive
<i>USH1G</i>	607696	Usher syndrome, type 1G, 606943 (3), Autosomal recessive
<i>USH2A</i>	608400	Usher syndrome, type 2A, 276901 (3), Autosomal recessive; Retinitis pigmentosa 39, 613809 (3)
<i>USP9X</i>	300072	Intellectual developmental disorder, X-linked 99, 300919 (3), X-linked recessive; Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968 (3), X-linked dominant
<i>VARS1</i>	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy, 617802 (3), Autosomal recessive
<i>VARS2</i>	612802	Combined oxidative phosphorylation deficiency 20, 615917 (3), Autosomal recessive
<i>VAX1</i>	604294	?Microphthalmia, syndromic 11, 614402 (3), Autosomal recessive
<i>VDR</i>	601769	Rickets, vitamin D-resistant, type IIA, 277440 (3), Autosomal recessive
<i>VHL</i>	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
<i>VIPAS39</i>	613401	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 (3), Autosomal recessive
<i>VKORC1</i>	608547	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3), Autosomal recessive; Warfarin resistance, 122700 (3), Autosomal dominant
<i>VLDLR</i>	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
<i>VMA21</i>	300913	Myopathy, X-linked, with excessive autophagy, 310440 (3), X-linked recessive

<i>VPS11</i>	608549	?Dystonia 32, 619637 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 12, 616683 (3), Autosomal recessive
<i>VPS13A</i>	605978	Choreoacanthocytosis, 200150 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS33B</i>	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogyriposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
<i>VPS37A</i>	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
<i>VPS45</i>	610035	Neutropenia, severe congenital, 5, autosomal recessive, 615285 (3), Autosomal recessive
<i>VPS53</i>	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>VSX2</i>	142993	Microphthalmia, isolated 2, 610093 (3); Microphthalmia with coloboma 3, 610092 (3)
<i>VWF</i>	613160	von Willebrand disease, type 1, 193400 (3), Autosomal dominant; von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 (3), Autosomal dominant, Autosomal recessive; von Willebrand disease, type 3, 277480 (3), Autosomal recessive
<i>WARS2</i>	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
<i>WAS</i>	300392	Wiskott-Aldrich syndrome, 301000 (3), X-linked recessive; Neutropenia, severe congenital, X-linked, 300299 (3), X-linked recessive; Thrombocytopenia, X-linked, intermittent, 313900 (3), X-linked recessive; Thrombocytopenia, X-linked, 313900 (3), X-linked recessive
<i>WASHC5</i>	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
<i>WDPCP</i>	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
<i>WDR19</i>	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
<i>WDR35</i>	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
<i>WDR45B</i>	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive

<i>WDR62</i>	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317 (3), Autosomal recessive
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
<i>WDR81</i>	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
<i>WFS1</i>	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
<i>WHRN</i>	607928	Deafness, autosomal recessive 31, 607084 (3), Autosomal recessive; Usher syndrome, type 2D, 611383 (3), Autosomal recessive
<i>WNK1</i>	605232	Neuropathy, hereditary sensory and autonomic, type II, 201300 (3), Autosomal recessive; Pseudohypoaldosteronism, type IIC, 614492 (3), Autosomal dominant
<i>WNT1</i>	164820	{Osteoporosis, early-onset, susceptibility to, autosomal dominant}, 615221 (3); Osteogenesis imperfecta, type XV, 615220 (3), Autosomal recessive
<i>WNT10A</i>	606268	Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal dominant, Autosomal recessive; Odontoonychodermal dysplasia, 257980 (3), Autosomal recessive
<i>WNT10B</i>	601906	Tooth agenesis, selective, 8, 617073 (3), Autosomal dominant; Split-hand/foot malformation 6, 225300 (3), Autosomal recessive
<i>WNT3</i>	165330	?Tetra-amelia syndrome 1, 273395 (3), Autosomal recessive
<i>WNT4</i>	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
<i>WNT7A</i>	601570	Fuhrmann syndrome, 228930 (3), Autosomal recessive; Ulna and fibula, absence of, with severe limb deficiency, 276820 (3), Autosomal recessive
<i>WRAP53</i>	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
<i>WRN</i>	604611	Werner syndrome, 277700 (3), Autosomal recessive
<i>WWOX</i>	605131	Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
<i>XDH</i>	607633	Xanthinuria, type I, 278300 (3), Autosomal recessive
<i>XIAP</i>	300079	Lymphoproliferative syndrome, X-linked, 2, 300635 (3), X-linked recessive
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPC</i>	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
<i>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive

<i>XRCC4</i>	194363	Short stature, microcephaly, and endocrine dysfunction, 616541 (3), Autosomal recessive
<i>XYLT1</i>	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
<i>XYLT2</i>	608125	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive; Spondyloocular syndrome, 605822 (3), Autosomal recessive
<i>YARS2</i>	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 (3), Autosomal recessive
<i>ZAP70</i>	176947	Immunodeficiency 48, 269840 (3), Autosomal recessive; Autoimmune disease, multisystem, infantile-onset, 2, 617006 (3), Autosomal recessive
<i>ZBTB24</i>	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2, 614069 (3), Autosomal recessive
<i>ZC4H2</i>	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
<i>ZDHHC9</i>	300646	Intellectual developmental disorder, X-linked syndromic, Raymond type, 300799 (3), X-linked
<i>ZFYVE26</i>	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
<i>ZIC3</i>	300265	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
<i>ZMPSTE24</i>	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive
<i>ZMYND10</i>	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
<i>ZNF335</i>	610827	Microcephaly 10, primary, autosomal recessive, 615095 (3), Autosomal recessive
<i>ZNF423</i>	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive
<i>ZNF469</i>	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive
<i>ZNF711</i>	314990	Intellectual developmental disorder, X-linked 97, 300803 (3), X-linked
<i>ZNHIT3</i>	604500	PEHO syndrome, 260565 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

OMIM release used for OMIM disease identifiers and descriptions: August 24, 2022

Possible phenotype mapping keys

- (1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
- (2) the disorder has been placed on the map by linkage; no mutation has been found
- (3) the molecular basis for the disorder is known; a mutation has been found in the gene
- (4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.